

Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
Kallikrein 3	KAK3	I
Kinectin	KTN1	G
Kinesin, heavy chain	KNSL1	G
Kinesin, light chain	KNS2	G
Kininogen, High molecular weight	KNG	I
Leptin	LEP	G
Leptin receptor	LEPR	G
Leukotriene A4 hydrolase		I
Leukotriene B4 receptor		I
Leukotriene C4 receptor		I
Leukotriene D4/E4 receptor		I
LH/choriogonadotropin (CG) receptor	LHCGR	G
LIM homeobox transcription factor 1, beta	LMX1B	G
Lipoprotein lipase	LPL	I
Lipoprotein receptor, Low Density	LDLR	T
Lipoxygenase 12 (platelets)	LOG12	I
Lipoxygenase 5 (leukocytes)		I
Low density lipoprotein receptor-related protein LRP precursor		T
Lysosomal acid lipase	LIPA	E
Malonyl CoA decarboxylase		E
Malonyl CoA transferase		E
Maltase-glucoamylase		E
Mannose binding protein	MBP	I
Mannosyl (alpha-1,6-)-glycoprotein beta-1, 2-	MGAT2	T
N-acetylglucosaminyltransferase		
MAPK kinase 1	MAPKK1; MEK1	G
MAPK kinase 4	MAPKK4; MEK4; SERK1	G
MAPK kinase 6	MAPKK6; MEK6	G
MAPKK kinase	MAPKKK	G
Matrix Gla protein	MGP	G
MEK kinase, MEKK		E

Melanocortin 2 receptor	MC2R	T
Melanocortin 4 receptor	MC4R	T
Methionine adenosyltransferase	MAT1A, MAT2A	E
Methionine synthase	MTR	E
Methionine synthase reductase	MTRR	E
Methylguanine-DNA methyltransferase	MGMT	E
Mevalonate kinase	MVK	E
MHC Class I: Tap1	ABCR, TAP1	I
MHC Class II: Tap2	TAP2, PSF2	I
Microphthalmia-associated transcription factor	MITF	G
Mismatch repair gene, PMSL1	PMS1	G
Mismatch repair gene, PMSL2	PMS2	G
Mitochondrial trifunctional protein, alpha subunit	HADHA	E
Mitochondrial trifunctional protein, beta subunit	HADHB	E
Mitogen-activated protein (MAP) kinase	MAPK	G
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Multidrug resistance associated protein	MRP	G
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Na <sup>+</sup> , K <sup>+</sup> ATPase, alpha	ATP1A1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 1	ATP1B1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 2	ATP1B2	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 3	ATP1B3	G
Na <sup>+</sup> /H <sup>+</sup> exchanger 1	NHE1	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 2	NHE2	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 3	NHE3	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 4	NHE4	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 5	NHE5	T
N-acetylgalactosamine-6-sulfate sulfatase	GALNS	E
N-acetylglucosamine-6-sulfatase	GNS	E
N-acetylglucosaminidase, alpha	NAGLU	E
N-acetyltransferase 1	NAT1	E
N-acetyltransferase 2	NAT2	E
N-acyl hydrolase		I
NADH dehydrogenase (ubiquinone) flavoprotein 1	NDUFV1	E
NADH-cytochrome b5 reductase	DIA1	E
NADPH-dependent cytochrome P450 reductase	POR	E
Nephrolithiasis 2	NPHL2	T
Nephronophthisis 2	NPHP2	T
Nephrosis 1	NPHS1	T
Neuroendocrine convertase 1	NEC1, PCSK1	E



Neurokinin A	NKNA	N
Neurokinin B	NKNB	N
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Niacin receptor		G
Niemann-Pick disease protein	NPC1	T
Nuclear factor kappa beta	NFKB	I
Nuclear factor of activated T cells (NFAT) complex, cytosolic	NFATC	G
Nuclear factor of activated T cells (NFAT) complex, preexisting component	NFATP	G
Nucleoside diphosphate kinase-A	NDPKA	E
Oncogene spi1		G
Opioid receptor, delta	OPRD1	N
Opioid receptor, kappa	OPRK1	N
Opioid receptor, mu	OPRM1	N
Ornithine transcarbamoylase	OTC, NME1	E
Osteoprotegerin	OPG	G
Otoferlin	OTOF	N
Oxytocin	OXT	N
Oxytocin receptor	OXTR	N
Paired-like homeodomain transcription factor 2	PITX2	G
Paired-like homeodomain transcription factor 3	PITX3	G
Paraoxonase PON1	PON1	E
Paraoxonase PON2	PON2	E
Paraoxonase PON3		E
Parathyroid hormone	PTH	G
Parathyroid hormone receptor	PTHr1	G
Parathyroid hormone related-peptide	PTHrP	G
Parathyroid hormone-like hormone	PTHrLH	G
Parvalbumin	PVALB	G
PCNA (proliferating cell nuclear antigen)		E
Peanut-like 1	PNUTL1	I
Peroxisomal membrane protein 1	PXMP1	S
Peroxisome biogenesis factor 1	PEX1	T
Peroxisome biogenesis factor 19	PEX19	T
Peroxisome biogenesis factor 6	PEX6	T
Peroxisome biogenesis factor 7	PEX7	T
Peroxisome proliferative activated receptor, alpha	PPARA	T
Peroxisome proliferative activated receptor, gamma	PPARG	T
P-glycoprotein 1	PGY1	T
P-glycoprotein 3	PGY3	T
Phenylethanolamine N-methyltransferase, PNMT	PNMT	E
Phosphodiesterase 1 / nucleotide	PDNP1	G

pyrophosphatase 1		
Phosphodiesterase 1 / nucleotide	PDNP2	G
pyrophosphatase 2		
Phosphodiesterase 1 / nucleotide	PDNP3	G
pyrophosphatase 3		
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C alpha		I
Phospholipase C beta		I
Phospholipase C delta	PLCD1	I
Phospholipase C epsilon		I
Phospholipase C gamma	PLCG1	I
Phosphomannomutase-2	PMM2	T
Phosphomannose isomerase-1, PMI1	MPI	T
Phosphoribosyl pyrophosphate synthetase	PRPS1	E
Pituitary adenylate cyclase activating peptide	PACAP	N
Pituitary adenylate cyclase activating peptide	PACAP1R	N
receptor		
Plasminogen activator, Tissue	PLAT; TPA	E
Platelet-activating factor receptor	PAFR	I
Plectin 1	PLEC1	T
Polycystin 1	PKD1	T
Polycystin 2	PKD2	T
Porphobilinogen deaminase	HMBS	E
Potassium channel, calcium-activated,	KCNN4	N
Potassium channel, subfamily K, member 1	KCNK1	N
Potassium channel, subfamily K, member 2	KCNK2	N
Potassium channel, subfamily K, member 3	KCNK3	N
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium inwardly-rectifying channel J11	KCNJ11	N
Potassium voltage-gated channel A1	KCNA1	N
Potassium voltage-gated channel E1	KCNE1	N
Potassium voltage-gated channel Q1	KCNQ1	N
Potassium voltage-gated channel Q2	KCNQ2	N
Potassium voltage-gated channel Q3	KCNQ3	N
POU domain, class 1, transcription factor 1 (Pit1)	POU1F1	G
POU domain, class 3, transcription factor 4	POU3F4	G
POU domain, class 4, transcription factor 3	POU4F3	G
Pre-B-cell leukemia transcription factor 1	PBX1	G
Preproglucagon	GCG;GLP1; GLP2	G
Progesterone receptor (RU486 binding	PGR	G

receptor)		
Prolactin	PRL	G
Prolactin receptor	PRLR	G
Proopiomelanocortin	POMC	N
Prostacyclin synthase		I
Prostaglandin 15-OH dehydrogenase	HGPD; PGDH	I
Prostaglandin D - DP receptor		I
Prostaglandin E1 receptor		I
Prostaglandin E2 receptor		I
Prostaglandin E3 receptor		I
Prostaglandin F - FP receptor		I
Prostaglandin F2 alpha receptor		I
Prostaglandin IP receptor		I
Prostaglandin-endoperoxidase synthase 2	PTGS2	G
Protease nexin 2	PN2	E
Protein C	PROC	I
Protein kinase DNA-activated	PRKDC	E
Protein S	PROS1	I
Pterin-4-alpha-carbinolamine	PCBD	
Purine nucleoside phosphorylase	NP	E
Purinergic receptor P1A1		N
Purinergic receptor P1A2		N
Purinergic receptor P1A3		N
Purinergic receptor P2X, 1	P2RX1	N
Purinergic receptor P2X, 2	P2RX2	N
Purinergic receptor P2X, 3	P2RX3	N
Purinergic receptor P2X, 4	P2RX4	N
Purinergic receptor P2X, 5	P2RX5	N
Purinergic receptor P2X, 6	P2RX6	N
Purinergic receptor P2X, 7	P2RX7	N
Purinergic receptor P2Y, 1	P2RY1	N
Purinergic receptor P2Y, 11	P2RY11	N
Purinergic receptor P2Y, 2	P2RY2	N
RAD51, DNA repair protein	RAD51	G
RAD52, DNA repair protein	RAD52	G
RAD54, DNA repair protein	RAD54	G
RAD55, DNA repair protein	RAD55	G
RAD57, DNA repair protein	RAD57	G
Recombination activating gene 1	RAG1	G
Recombination activating gene 2	RAG2	G
Red cone pigment	RCP	S
Replication factor A		E
Replication factor C	RFC2	E
Retinaldehyde binding protein 1	RLBP1	T
Retinoic acid receptor, alpha	RARA	G
Retinoic acid receptor, beta	RARB	G
Retinoic acid receptor, gamma	RARG	G
Retinoid X receptor, alpha	RXRA	G

Retinoid X receptor, beta	RXRB	G
Retinoid X receptor, gamma	RXRG	G
Retinol binding protein 1		T
Retinol binding protein 2		T
Retinol binding protein 4	RBP4	T
Ribonucleotide reductase, RRM		E
Ribosephosphate pyrophosphokinase		E
Ribosomal protein L13A	RPL13A	G
Ribosomal protein S19	RPS19	E
Ribosomal protein S4, X-linked	RPS4X	E
Ribosomal protein S6 kinase	RPS6KA3	E
Ribosomal protein S9	RPS9	G
S-adenosylmethionine decarboxylase, AMD		E
Secretin	SCT	T
Secretin receptor, SCTR	SCTR	T
Serine hydroxymethyltransferase	SHMT	E
Serotonin N-acetyltransferase	SNAT	E
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Slug protein		G
Small nuclear ribonucleoprotein polypeptide N	SNRPN	S
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage gated, type IV, alpha	SCN4A	N
polypeptide		
Sodium channel, voltage gated, type V, alpha	SCN5A	N
polypeptide		
Sodium channel, voltage-gated, type 1, beta	SCN1B	N
polypeptide		
Solute carrier family 1 (amino acid transporter), member 6	SLC1A6	T
Solute carrier family 1 (glial high affinity glutamate transporter), member 3	SLC1A3	T
Solute carrier family 1 (glutamate transporter), member 1	SLC1A1	T

Solute carrier family 1 (glutamate transporter), member 2	SLC1A2	T
Solute carrier family 1 (neutral amino acid transporter), member 4	SLC1A4	T
Solute carrier family 10 (sodium/bile acid cotransporter family), member 1	SLC10A1	T
Solute carrier family 10 (sodium/bile acid cotransporter family), member 2	SLC10A2	T
Solute carrier family 12, member 1	SLC12A1	T
Solute carrier family 12, member 2	SLC12A2	T
Solute carrier family 12, member 3	SLC12A3	T
Solute carrier family 14, member 2	SLC14A2	T
Solute carrier family 15 (H <sup>+</sup> /peptide transporter, intestinal), member 1	SLC15A1	T
Solute carrier family 15 (H <sup>+</sup> /peptide transporter, kidney), member 2	SLC15A2	T
Solute carrier family 16 (monocarboxylate transporter), member 1	SLC16A1	T
Solute carrier family 16 (monocarboxylate transporter), member 7	SLC16A7	T
Solute carrier family 17, member 1	SLC17A1	T
Solute carrier family 17, member 2	SLC17A2	T
Solute carrier family 18, member 3	SLC18A3	T
Solute carrier family 19 (folate transporter), member 1	SLC19A1	T
Solute carrier family 2 (facilitated glucose transporter), member 1	SLC2A1	T
Solute carrier family 2 (facilitated glucose transporter), member 2	SLC2A2	T
Solute carrier family 2 (facilitated glucose transporter), member 3	SLC2A3	T
Solute carrier family 2 (facilitated glucose transporter), member 4	SLC2A4	T
Solute carrier family 2 (facilitated glucose transporter), member 5	SLC2A5	T
Solute carrier family 20, member 1	SLC20A1	T
Solute carrier family 20, member 2	SLC20A2	T
Solute carrier family 20, member 3	SLC20A3	T
Solute carrier family 21, member 2	SLC21A2	T
Solute carrier family 21, member 3	SLC21A3	T
Solute carrier family 22, member 1	SLC22A1	T
Solute carrier family 22, member 2	SLC22A2	T
Solute carrier family 22, member 5	SLC22A5	T
Solute carrier family 25, member 12	SLC25A12	T
Solute carrier family 3 (facilitated glucose transporter), member 1	SLC3A1	T
Solute carrier family 4 (anion exchanger), member 1	SLC4A1	T

Solute carrier family 4 (anion exchanger), member 2	SLC4A2	T
Solute carrier family 4 (anion exchanger), member 3	SLC4A3	T
Solute carrier family 5 (sodium/glucose transporter), member 1	SLC5A1	T
Solute carrier family 5 (sodium/glucose transporter), member 2	SLC5A2	T
Solute carrier family 5 (sodium/glucose transporter), member 5	SLC5A5	T
Solute carrier family 5, member 3	SLC5A3	T
Solute carrier family 6 (GAMMA-AMINOBUTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Solute carrier family 6 (neurotransmitter transporter, serotonin), member 4	SLC6A4	T
Solute carrier family 6, member 10	SLC6A10	T
Solute carrier family 6, member 6	SLC6A6	T
Solute carrier family 6, member 8	SLC6A8	T
Solute carrier family 7(amino acid transporter), member 1	SLC7A1	T
Solute carrier family 7(amino acid transporter), member 2	SLC7A2	T
Solute carrier family 7(amino acid transporter), member 7	SLC7A7	T
Solute carrier family 8 (sodium/calcium exchanger), member 1	SLC8A1	T
Somatostatin	SST	N
Somatostatin receptor, SSTR1	SSTR1	N
Somatostatin receptor, SSTR2	SSTR2	G
Somatostatin receptor, SSTR3	SSTR3	N
Somatostatin receptor, SSTR4	SSTR4	N
Somatostatin receptor, SSTR5	SSTR5	N
Sorcin	SRI	T
SOS1 guanine nucleotide exchange factor	SOS1	G
Steroid 5 alpha reductase 1	SRD5A1	E
Steroid 5 alpha reductase 2	SRD5A2	E
Steroid hormone receptor responsive DNA elements		G
Sterol carrier protein 2	SCP2	T
Succinic semi-aldehyde dehydrogenase	ssadh	E
Sucrase		E
Sulfonylurea receptor	SUR	G
Synaptic vesicle amine transporter	SVAT	N
Tachykinin receptor, NK1R	TACR1	N

Tachykinin receptor, NK2R	TACR2	N
Tachykinin receptor, NK3R	TACR3	N
Terminal deoxynucleotidyltransferase	TDT	I
Thiopurine S-methyltransferase	TPMT	E
Thrombopoietin	THPO	G
Thromboxane A synthase 1	TBXAS1	I
Thromboxane A2	TXA2	I
Thromboxane A2 receptor	TBXA2R	I
Thymidylate synthase	TYMS	E
Thymopoietin	TMPO	G
Thyroid hormone receptor, beta	THRB	G
Thyroid-stimulating hormone receptor	TSHR	G
Thyroid-stimulating hormone, alpha	TSHA	G
Thyroid-stimulating hormone, beta	TSHB	G
Topoisomerase I		E
Topoisomerase II		E
Transcription factor 1, hepatic	TCF1	G
Transcription factor 2, hepatic	TCF2	G
Transcription factor 3	TCF3	G
Transcription factor binding to IGHM enhancer 3	TFE3	G
Transcription factor, TUPLE1	TUPLE1	N
Transcription termination factor, RNA polymerase 1	TTF1	G
Transcription termination factor, RNA polymerase 2	TTF2	G
Transcription termination factor, RNA polymerase 3	TTF3	G
Transferrin	TF	G
Transferrin receptor	TFRC	G
Transthyretin	TTR	T
Tubulin		S
Tumour necrosis factor (TNF) receptor associated factor 1	TRAF1	I
Tumour necrosis factor (TNF) receptor associated factor 2	TRAF2	I
Tumour necrosis factor (TNF) receptor associated factor 3	TRAF3	I
Tumour necrosis factor (TNF) receptor associated factor 4	TRAF4	I
Tumour necrosis factor (TNF) receptor associated factor 5	TRAF5	I
Tumour necrosis factor (TNF) receptor associated factor 6	TRAF6	I
Tumour necrosis factor alpha	TNFA	I
Tumour necrosis factor alpha receptor	TNFAR	I
Tumour necrosis factor beta	TNFB	I
Tumour necrosis factor beta receptor	TNFBR	I

Tumour protein p53	TP53, P53	G
Tumour protein p63	TP63	G
Tumour suppressor gene DRA	DRA	I
Ubiquitin		G
Ubiquitin activating enzyme, E1		E
Ubiquitin B	UBB	G
Ubiquitin C	UBC	G
Ubiquitin carboxyl-terminal esterase L1	UCHL1	G
Ubiquitin protein ligase E3A	UBE3A	E
UDP-glucose pyrophosphorylase		E
UDP-glucuronosyltransferase 1	ugt1d, UGT1	E
UDP-glucuronosyltransferase 2	UGT2	E
Uncoupling protein 1		T
Uncoupling protein 3	UCP3	T
Uridine monophosphate kinase	UMPK	I
Uridine monophosphate synthetase	UMPS	I
Uridinediphosphate(UDP)-galactose-4-epimerase	GALE	E
Vimentin	VIM	I
Vitamin B12-binding (R) protein		G
Vitamin D receptor	VDR	G
Xanthine dehydrogenase	XDH	E
Xeroderma pigmentosum, complementation group A	XPA	E
Xeroderma pigmentosum, complementation group B	XPB	E
Xeroderma pigmentosum, complementation group C	XPC	E
Xeroderma pigmentosum, complementation group D		E
Xeroderma pigmentosum, complementation group E		E
Xeroderma pigmentosum, complementation group F	XPF	E
Xeroderma pigmentosum, complementation group G	ERCC5	E
X-ray repair gene	XRCC9	G
Xylitol dehydrogenase		E
YY1 transcription factor	YY1	G

2. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim



- 1.
3. A set according to claim 1 or 2 in which a minority of said probes for listed genes are absent.
4. A set according to claim 1 or 2 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.
5. A set according to claim 1 or 2 in which a limited number of probes are replaced by probes for non-listed genes.
6. A set of probes for a core group of genes according to any of claims 1 to 5 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.
7. A set according to any of claims 1 to 6 consisting of probes for members of a sub-group of the core group.
8. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.
9. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.
10. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.
11. A set according to claim 8 or 9 in which said substrate is a semiconductor microchip.
12. A set according to any preceding claim for use in a biological assay for detection of said gene variants.
13. A set according to any preceding claim for use in the measurement of differential gene expression levels.
14. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
15. A medical device including a set according to any of claims 1 to 13 for use in an array for detection of differential gene expression levels.
16. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 1) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 1 and 3 to 13 and relating the probe hybridisation pattern to said variations.
17. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 2) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 2 to 13 and relating the probe interaction pattern to said variations.
18. Use of a set or device according to any of claims 1 to 13 for the prognosis and management of patients suffering from or at risk of adverse events.

19. Use of a set or device according to any of claims 1 to 13 for predicting likely therapeutic response and adverse events following therapeutic intervention.
20. Use of a set or device according to any of claims 1 to 13 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
20. Use of a set or device according to any of claims 1 to 13 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
21. Use of a set or device according to any of claims 1 to 13 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
22. Use of a set or device according to any of claims 1 to 13 for the development of new strategies of therapeutic intervention and in clinical trials.
23. Use of a set or device according to any of claims 1 to 13 for construction of and generation of algorithms for patient and healthcare management.
24. Use of a set or device according to any of claims 1 to 13 for modelling or assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations
25. Use of a set or device according to any of claims 1 to 13 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
26. Use of a set or device according to any of claims 1 to 13 for predicting optimum configuration/management of thereapeutic intervention.
27. A method according to claim 16 or 17 in which the identification of gene variants is indicative of a higher risk of experiencing adverse events for the patient or individual.
28. A method for generating a model to assess whether a patient or individual or population or group is or are likely to experience adverse events, which method comprises:
  - i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from adverse events;
  - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the adverse events;
  - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 1 to 7;
  - iv) calculating the frequencies of these alleles in the samples from i) and ii);
  - v) comparing the frequencies of these alleles in i) and ii);
  - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of experiencing adverse events.
29. A method for assessing whether a given subject will be at risk of developing symptoms, which comprises comparing said subject's genotype with a model generated by the method of claim 28.
30. A method according to any of claims 16, 17, 28 and 29 wherein at least one step is computer-controlled.
31. An assay suitable for use in a method according to any of claims 16, 17, 28 and 29; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 1 to 7 in a biological sample.

32. A formatted assay technique (kit) for use in assessing the risk of a patient or individual experiencing adverse events; said kit comprising:
- i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 1 or 3 to 7 in a sample of human DNA;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual experiencing adverse events.
33. A formatted assay technique (kit) for use in assessing the risk of a patient or individual experiencing adverse events; said kit comprising:
- i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core group of genes as defined in any of claims 2 to 7 in an expressed-protein-containing human sample;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual experiencing adverse events.
34. A set of probes according to claim 1, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.
35. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to cancer; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

#### KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME  
 T TRANSPORT & STORAGE  
 S STRUCTURAL  
 I IMMUNITY  
 N NERVOUS TRANSMISSION  
 G GROWTH & DIFFERENTIATION

#### ONCOLOGY GENE LIST

	HUGO gene symbol	Protein function
Absent in melanoma 1 gene	AIM1	G
Actin, alpha, cardiac	ACTC	S
Actin, alpha, skeletal	ACTA1	S
Actin, alpha, smooth, aortic	ACTA2	S
Activin		G
Activin A receptor, type 2B	ACVR2B	G
Activin A receptor, type 2-like kinase 1	ACVRL1	G
Adenomatous polyposis coli tumour suppressor	APC	G

gene		
Adenosine deaminase	ADA	E
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N
Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenyl cyclase		N
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
Adrenocorticotrophic hormone (ACTH) receptor	ACTHR	G
Albumin, ALB	ALB	T
Alcohol dehydrogenase 3	ADH3	E
Aldehyde dehydrogenase 1	ALDH1	E
Aldehyde dehydrogenase 10	ALDH10	E
Aldehyde dehydrogenase 2	ALDH2	E
Aldehyde dehydrogenase 5	ALDH5	E
Aldehyde dehydrogenase 6	ALDH6	E
Aldehyde dehydrogenase 7	ALDH7	E
Aldosterone receptor	MLR	G
alpha tectorin	TECTA	G
alpha1-antitrypsin	PI	E
alpha-actinin 2	ACTN2	G
alpha-actinin 3	ACTN3	G
Alpha-fetoprotein	AFP	G
alpha-synuclein	SNCA	N
Amphiregulin	AREG	G
Amyloid beta A4 precursor protein	APP	N
Amyloid beta A4 precursor-like protein	APLP	N
Androgen receptor	AR	G
Angiopoietin 1	ANGPT1	G
Angiopoietin 2	ANGPT2	G
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensin receptor 1	AGTR1	T
Angiotensin receptor 2	AGTR2	T
Angiotensinogen	AGT	E

Annexin 1	ANX 1	I
Antidiuretic hormone receptor	ADHR	T
Antithrombin III	AT3	E
AP-2, alpha	TFAP2A	G
AP-2, beta	TFAP2B	G
AP-2, gamma	TFAP2C	G
Apaf-1		S
Apoptosis antigen 1	APT1	I
Apoptosis antigen ligand 1	APT1LG1	I
Apoptosis-inducing factor	AIF	I
Apurinic endonuclease	APE	E
Arginine vasopressin	AVP	N
Arginosuccinate synthetase	ASS	E
Aryl hydrocarbon receptor	AHR	T
Aryl hydrocarbon receptor nuclear translocator	ARNT	T
Asparagine synthetase	AS	E
Aspartate receptor		N
Ataxia telangiectasia complementation group D	ATD, ATDC	G
Ataxia telangiectasia gene, AT	ATM	G
ATP cobalamin adenosyltransferase		E
ATP sulphurylase	atpsk2	E
ATP-binding cassette transporter 7	ABC7	I
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G
Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
Atrophin 1	DRPLA	G
Bagpipe homeobox, drosophila homolog of, 1	BAPX1	G
B-cell CLL/lymphoma 1	BCL1	I
B-cell CLL/lymphoma 10	BCL10	I
B-cell CLL/lymphoma 3	BCL3	I
B-cell CLL/lymphoma 4	BCL4	I
B-cell CLL/lymphoma 5	BCL5	I
B-cell CLL/lymphoma 6	BCL6	I
B-cell CLL/lymphoma 7	BCL7	I
B-cell CLL/lymphoma 8	BCL8	I
B-cell CLL/lymphoma 9	BCL9	I
BCL2-associated X protein	BAX	G
BCL2-related protein A1	BCL2A1	G
Beckwith-Wiedemann region 1A	BWR1A	G
Benzodiazepine receptor		N
beta 2 microglobulin	B2M	I
beta-endorphin receptor		N
beta-synuclein	SNCB	N
Bleomycin hydrolase	BLMH	E
Bone morphogenetic protein, BMP1	BMP1	G
Bone morphogenetic protein, BMP2	BMP2	G
Bone morphogenetic protein, BMP3	BMP3	G

Bone morphogenetic protein, BMP4	BMP4	G
Bone morphogenetic protein, BMP5	BMP5	G
Bone morphogenetic protein, BMP6	BMP6	G
Bone morphogenetic protein, BMP7	BMP7	G
Bone morphogenetic protein, BMP8	BMP8	G
Bradykinin receptor B1		I
Bradykinin receptor B2		I
Brain derived neurotrophic factor	BDNF	G
Brain derived neurotrophic factor (BDNF) receptor	BDNFR	G
Branched chain aminotransferase 1, cytosolic	BCAT1	E
Branched chain aminotransferase 2, mitochondrial	BCAT2	E
BRCA1-associated RING domain gene 1	BARD1	G
Breakpoint cluster region	BCR	G
Breast cancer 1	BRCA1	G
Breast cancer 2	BRCA2	G
Breast cancer, ductal, 1	BRCD1	G
Breast cancer, ductal, 2	BRCD2	G
Bruton agammaglobulinaemia tyrosine kinase	BTK	G
C1 inhibitor		E
Cadherin E	CDH1	G
Cadherin EP		G
Cadherin N	CDH2	G
Cadherin P	CDH3	G
Calbindin 1	CALB1	G
Calbindin D9K	CALB3	G
Calcitonin receptor /Calcitonin gene-related peptide receptor	CALCR	N
Calcitonin/Calcitonin gene-related peptide alpha	CALCA	N
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha-1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha-1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha-1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha-1E (CACNL1A6)	CACNA1E	N
Calcium channel, voltage-dependent, Alpha-2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, L type, alpha 1S subunit	CACNA1S	N
Calcium channel, voltage-dependent,	CACNG2	N

## Neuronal, Gamma

Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	CACNA1A	N
Calcium channel, voltage-dependent, T-type		N
Calmodulin 1	CALM1	G
Calmodulin 2	CALM2	G
Calmodulin 3	CALM3	G
Calmodulin-dependant protein kinase II	CAMK2A	G
Calnexin	CANX	G
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Cardiac-specific homeobox, CSX	CSX	G
Cartilage-hair hypoplasia gene	CHH	N
Caspase 1	CASP1	G
Caspase 10	CASP10	G
Caspase 2	CASP2	G
Caspase 3	CASP3	G
Caspase 4	CASP4	G
Caspase 5	CASP5	G
Caspase 6	CASP6	G
Caspase 7	CASP7	G
Caspase 8	CASP8	G
Caspase 9	CASP9	G
Catenin, beta	CTNNB1	G
CD1	CD1	I
CD10	CD10	I
CD4	CD4	I
CEA		G
Cell adhesion molecule, intercellular, ICAM	ICAM1	G
Cell adhesion molecule, leukocyte-endothelial, LECAM (CD62)	LECAM1	G
Cell adhesion molecule, liver, LCAM	LCAM	G
Cell adhesion molecule, neural, NCAM1	NCAM1	G
Cell adhesion molecule, neural, NCAM120	NCAM120	G
Cell adhesion molecule, neural, NCAM2	NCAM2	G
Cell adhesion molecule, platelet-endothelial, PECAM	PECAM1	G
Cell adhesion molecule, vascular, VCAM	VCAM1	G
c-erbB1	ERBB1	G
c-erbB2	ERBB2	G
c-erbB3	ERBB3	G
c-erbB4	ERBB4	G
Ceruloplasmin precursor	CP	E
Chemokine receptor CXCR1	CXCR1	I
Chemokine receptor CXCR2	CXCR2	I
Cholecystokinin	CCK	N

Cholecystikinin B receptor	CCKBR	N
Ciliary neurotrophic factor (CNTF)	CNTF	G
Ciliary neurotrophic factor (CNTF) receptor	CNTFR	G
c-kit receptor tyrosine kinase		G
Clathrin		T
Clusterin	CLU	G
Collagen IV alpha 4	COL4A4	S
Collagen IV alpha 5	COL4A5	S
Collagen IV alpha 6	COL4A6	S
Colony-stimulating factor 1	CSF1	G
Colony-stimulating factor 1 receptor	CSF1R	G
Colony-stimulating factor 2	CSF2	G
Colony-stimulating factor 2 alpha receptor	CSF2RA	G
Colony-stimulating factor 2 beta receptor	CSF2RB	G
Colony-stimulating factor 3	CSF3	G
Colony-stimulating factor 3 receptor	CSF3R	G
Complement component C1 inhibitor	C1NH	I
Complement component C1qa	C1QA	I
Complement component C1qb	C1QB	I
Complement component C1qg	C1QG	I
Complement component C1r	C1R	I
Complement component C1s	C1S	I
Complement component C2	C2	I
Complement component C3	C3	I
Complement component C4A	C4A	I
Complement component C4B	C4B	I
Complement component C5	C5	I
Complement component C6	C6	I
Complement component C7	C7	I
Complement component C8	C8B	I
Complement component C9	C9	I
Complex III		E
Core-binding factor, alpha 1	CBFA1	G
Core-binding factor, alpha 2	CBFA2	G
Core-binding factor, beta	CBFB	G
Corticotrophin-releasing hormone	CRH	T
Corticotrophin-releasing hormone receptor	CRHR1	T
c-src tyrosine kinase	CSK	G
Cyclic AMP-dependent protein kinase	PKA	E
Cyclin A	CCNA	G
Cyclin B	CCNB	G
Cyclin C	CCNC	G
Cyclin D	CCND1	G
Cyclin E	CCNE	G
Cyclin F	CCNF	G
Cyclin-dependent kinase 1	CDK1	G
Cyclin-dependent kinase 10	CDK10	G
Cyclin-dependent kinase 2	CDK2	G



CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystathionase	CTH	E
Cystathione beta synthase	CBS	E
Cystic fibrosis transmembrane conductance regulator, CFTR	CFTR	N
Cytidine deaminase	CDA	E
Cytidine-5-prime-triphosphate synthetase	CTPS	E
Cytochrome a		E
Cytochrome c		E
Cytochrome c oxidase, MTCO		E
Cytokine-suppressive antiinflammatory drug- binding protein 1	CSBP1	I
Cytokine-suppressive antiinflammatory drug- binding protein 2	CSBP2	I
Defender against cell death 1	DAD1	G
Deleted in colorectal carcinoma	DCC	G
Deleted in malignant brain tumours 1	DMBT1	G
Deoxycytidine kinase DCK		E
Deoxyuridine triphosphatase; dUTPase		E
Desert hedgehog, dhh		G
Dihydrofolate reductase	DHFR	E
Dihydrolipoyl dehydrogenase		E
Dihydropyrimidine dehydrogenase	DPYD	E
DM-Kinase	DMPK	E
DNA damage binding protein, DDB1	DDB1	S
DNA damage binding protein, DDB2	DDB2	S
DNA directed polymerase, alpha	POLA	E
DNA glycosylases		E
DNA helicases		E
DNA Ligase 1	LIG1	E
DNA methyltransferase	DNMT	E
DNA polymerase 1		E
DNA polymerase 2		E
DNA polymerase 3		E
DNA primase		E
DNA-damage-inducible transcript 3	DDIT3	S
DNA-dependant RNA polymerase		E
DOPA decarboxylase	DDC	E
Dopamine receptors D1	DRD1	N
Dopamine receptors D2	DRD2	N
Dopamine receptors D3	DRD3	N
Dopamine receptors D4	DRD4	N

Dopamine receptors D5	DRD5	N
Dynamin	DNM1	G
Dynorphin receptor		N
Dysferlin	DYS, DYSF	E
Dyskerin	DKC1	S
EB1		G
Endoglin	ENG	S
Endothelin 1	EDN1	N
Endothelin 2	EDN2	N
Endothelin 3	EDN3	N
Endothelin converting enzyme	ECE1	N
Endothelin receptor type A	EDNRA	N
Endothelin receptor type B	EDNRB	N
Enolase	ENO1	E
Ephrin receptor tyrosine kinase A	EPHA	G
Ephrin receptor tyrosine kinase B	EPHB	G
Epidermal growth factor	EGF	G
Epidermal growth factor receptor	EGFR	G
Estrogen receptor	ESR	G
Eukaryotic initiation translation factor	EIF4E	G
EWS RNA-binding protein	EWSR1	G
Excision repair complementation group 1 protein	ERCC1	E
Excision repair complementation group 2 protein	ERCC2	E
Excision repair complementation group 2 protein	ERCC3	E
Excision repair complementation group 4 protein	ERCC4	E
Excision repair complementation group 6 protein	ERCC6	E
Exostosin 1	EXT1	S
Exostosin 2	EXT2	S
FADH dehydrogenase		E
Fanconi anemia, complementation group C	FANCC	T
Fanconi anemia, complementation group D	FANCD	T
Fc fragment of IgG, high affinity IA, receptor for	FCGR1A	G
Fc fragment of IgG, low affinity IIa, receptor for	FCGR2A	G
(CD32)		
Fc fragment of IgG, low affinity IIIa, receptor for	FCGR3A	G
(CD16)		
Ferrochelatase	FECH	E
Fibrillin 1	FBN1	G
Fibroblast growth factor	FGF1	G
Fibroblast growth factor receptor 1	FGFR1	G
Fibroblast growth factor receptor 2	FGFR2	G
Fibroblast growth factor receptor 3	FGFR3	G
Fibronectin precursor	FN1	G

Folic acid receptor	FOLR	G
Follicle stimulating hormone receptor	FSHR, ODG1	G
Follicle stimulating hormone, FSH	FSHB	G
Follicular lymphoma variant translocation 1	FVT1	I
Forkhead rhabdomyosarcoma gene	FKHR	G
Forkhead transcription factor 14	FKHL14	G
Forkhead transcription factor 7	FKHL7	G
Fucosyltransferase 2	FUT2	T
Fucosyltransferase 3	FUT3	T
G/T mismatch binding protein	GTBP, MSH6	G
GABA receptor, alpha 1	GABRA1	N
GABA receptor, alpha 2	GABRA2	N
GABA receptor, alpha 3	GABRA3	N
GABA receptor, alpha 4	GABRA4	N
GABA receptor, alpha 5	GABRA5	N
GABA receptor, alpha 6	GABRA6	N
GABA receptor, beta 1	GABRB1	N
GABA receptor, beta 2	GABRB2	N
GABA receptor, beta 3	GABRB3	N
GABA receptor, gamma 1	GABRG1	N
GABA receptor, gamma 2	GABRG2	N
GABA receptor, gamma 3	GABRG3	N
Gadd45 (growth arrest & DNA-damage-inducible protein)		E
Galactosyltransferase 1	GT1	G
Galactosyltransferase, alpha 1,3	GGTA1	G
Galactosyltransferase, beta 3	B3GALT	G
Gastrin	GAS	G
Gastrin releasing peptide	GRP	T
Glioma chloride ion channel, GCC		G
Glucagon receptor	GCGR	G
Glucagon synthase		T
Glucocorticoid receptor	GRL	G
Glutamate receptor 1	GLUR1	N
Glutamate receptor 2	GLUR2	N
Glutamate receptor 3	GLUR3	N
Glutamate receptor 4	GLUR4	N
Glutamate receptor 5	GLUR5	N
Glutamate receptor 6	GLUR6	N
Glutamate receptor 7	GLUR7	N
Glutamate receptor, ionotropic, NMDA 1	NMDAR1	N
Glutamate receptor, ionotropic, NMDA 2A	NMDAR2A	N
Glutamate receptor, ionotropic, NMDA 2B	NMDAR2B	N
Glutamate receptor, ionotropic, NMDA 2C	NMDAR2C	N
Glutamate receptor, ionotropic, NMDA 2D	NMDAR2D	N
Glutathione	GSH	T
Glutathione S-transferase mu 1, GSTM1	GSTM1	E
Glutathione S-transferase theta 1, GSTT1	GSTT1	E
Glutathione S-transferase, GSTZ1	GSTZ1	E

Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
GAPDH		
Glycerol kinase	GK	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
Glycine receptor, alpha	GLRA2	N
Glycine receptor, beta		N
Glycine transporter	GLYT	N
Glypican 3	GPC3, SDYS	G
Gonadotropin releasing hormone	GNRH	G
Gonadotropin releasing hormone receptor	GNRHR	G
Growth factor receptor-bound protein 2	GRB2	G
Growth hormone 1	GH1	G
Growth hormone 2 (placental)	GH2	G
Growth hormone receptor	GHR	G
Growth hormone releasing hormone (GHRH)	GHRH	G
Growth hormone releasing hormone receptor	GHRHR	G
Growth/differentiation factor 5	GDF5	G
Growth-regulated protein precursor, GRO	GRO	I
GTPase-activating protein, GAP	RASA1	G
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 1, GNAI1	GNAI1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 2, GNAI2	GNAI2	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 3, GNAI3	GNAI3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS1	GNAS1	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS2	GNAS2	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS3	GNAS3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS4	GNAS4	N
Guanine nucleotide-binding protein, q polypeptide	GNAQ	N
Guanylate kinase		E
H(+), K(+) - ATPase	ATP4B	N
Hairless	HR	G
Hela tumor suppression gene	HTS1	G
Heparin binding epidermal growth factor	HBEGF	G
Hepatitis B virus integration site 1	HVBS1	I
Hepatitis B virus integration site 2	HVBS6	I
High mobility group protein C	HMGIC	G
High mobility group protein Y	HMG1Y	G
Histamine receptors, H1		N
Histamine receptors, H2		N
Histamine receptors, H3		N

HLH transcription factor HAND1	HAND1	G
HLH transcription factor HAND2	HAND2	G
HMG-CoA reductase	HMGCR	E
HMG-CoA synthase	HMGCS2	E
Homeobox (HOX) gene A13	HOXA13	G
Homeobox 11	HOX11	G
Homeobox HB24	HLX1	G
Homogentisate 1,2 dioxygenase	HGD	E
Hormone-sensitive lipase	HSL	E
HSSB, replication protein		E
Human placental lactogen	CSH1	G
Ibonucleoside diphosphate reductase		E
Ikaros gene	IKAROS	G
Inhibin, alpha	INHA	G
Inhibin, beta A	INHBA	G
Inhibin, beta B	INHBB	G
Inhibin, beta C	INHBC	G
Inositol 1,4,5-triphosphate receptor 3	ITPR3	G
Insulin	INS	G
Insulin receptor	INSR	G
Insulin-like growth factor 1	IGF1	G
Insulin-like growth factor 1 receptor	IGF1R	G
Insulin-like growth factor 2	IGF2	G
Insulin-like growth factor 2 receptor	IGF2R	G
Integrin beta 1	ITGB1	G
Integrin beta 2	ITGB2	G
Integrin beta 3	ITGB3	G
Integrin beta 4	ITGB4	G
Integrin beta 5	ITGB5	G
Integrin beta 6	ITGB6	G
Integrin beta 7	ITGB7	G
Integrin, alpha 1	ITGA1	G
Integrin, alpha 2	ITGA2	G
Integrin, alpha 4	ITGA4	G
Integrin, alpha 5	ITGA5	G
Integrin, alpha 6	ITGA6	G
Integrin, alpha M	ITGAM	G
Interferon alpha	IFNA1	I
Interferon beta	IFNB	I
Interferon gamma	IFNG	I
Interferon gamma receptor 1	IFNGR1	I
Interferon gamma receptor 2	IFNGR2	I
Interferon regulatory factor 1	IRF1	I
Interferon regulatory factor 4	IRF4	I
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) 10	IL10	I

Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I
Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
Janus kinase 1	JAK1	G
Janus kinase 2	JAK2	G
Janus kinase 3	JAK3	G
Laminin 5, alpha 3	LAMA3	G
Laminin 5, beta 3	LAMB3	G
Laminin 5, gamma 2	LAMC2	G
Laminin M	LAMM	G
Laminin receptor 1	LAMR1	G
Latent transforming growth factor-beta binding protein 2	LTBP2	G
Leptin	LEP	G
Leptin receptor	LEPR	G
Leukaemia inhibitory factor	LIF	G
Leukaemia inhibitory factor receptor	LIFR	G
Leukotriene A4 hydrolase		I
Leukotriene B4 receptor		I
Leukotriene C4 receptor		I
Leukotriene D4/E4 receptor		I
LH/choriogonadotropin (CG) receptor	LHCGR	G
LIM homeobox protein 1	LHX1	G
LIM homeobox protein 2	LHX2	G
LIM homeobox protein 3	LHX3	G
LIM homeobox protein 4	LHX4	G

Limbic associated membrane protein	LAMP	G
LIM-domain only protein 1	LMO1	G
LIM-domain only protein 2	LMO2	G
LIM-domain only protein 3	LMO3	G
LIM-domain only protein 4	LMO4	G
Lipoma-preferred partner gene	LPP	G
Lipoxygenase 12 (platelets)	LOG12	I
Lipoxygenase 5 (leukocytes)		I
Long QT-type 2 potassium channels	LQT2, KCNH2	T
Lowe oculocerbrorenal syndrome gene	OCRL	E
Luteinizing hormone-releasing hormone		N
Luteinizing hormone-releasing hormone receptor		N
Lymphoblastic leukemia derived sequence 1	LYL1	I
Lymphocyte-specific protein tyrosine kinase	LCK	I
Lymphoid enhancer-binding factor	LEF-1	G
Macrophage activating factor	MAF	I
MAD (mothers against decapentaplegic, Drosophila) homologue 3	MADH3	G
MAD (mothers against decapentaplegic, Drosophila) homologue 4	MADH4	G
MADS box transcription-enhancer factor 2A	MEF2A	G
MADS box transcription-enhancer factor 2B	MEF2B	G
MADS box transcription-enhancer factor 2C	MEF2C	G
MADS box transcription-enhancer factor 2D	MEF2D	G
Malignant proliferation, eosinophil gene	MPE	I
MAPK kinase 1	MAPKK1; MEK1	G
MAPK kinase 4	MAPKK4; MEK4; SERK1	G
MAPK kinase 6	MAPKK6; MEK6	G
MAPKK kinase	MAPKKK	G
MAX-interacting protein 1	MXI1	G
MEK kinase, MEKK		E
Melanocortin 1 receptor	MC1R	T
Menin	MEN1	G
Methionine adenosyltransferase	MAT1A, MAT2A	E
Methionine synthase	MTR	E
Methionine synthase reductase	MTRR	E
Methylguanine-DNA methyltransferase	MGMT	E
MHC Class I: A		I
MHC Class I: B		I
MHC Class I: C		I
MHC Class I: LMP-2, LMP-7		I
MHC Class I: Tap1	ABCR, TAP1	I
MHC Class II: DP	HLA-DPB1	I
MHC Class II: DQ		I
MHC Class II: DR		I
MHC Class II: Tap2	TAP2, PSF2	I

MHC Class II:Complementation group A	MHC2TA	I
MHC Class II:Complementation group B	rfxank	I
MHC Class II:Complementation group C	RFX5	I
MHC Class II:Complementation group D	RFXAP	I
Midline 1	MID1	G
Mismatch repair gene, PMSL1	PMS1	G
Mismatch repair gene, PMSL2	PMS2	G
Mitogen-activated protein (MAP) kinase	MAPK	G
Motilin	MLN	G
Msh homeobox homolog 1	MSX1	G
Msh homeobox homolog 2	MSX2	G
Mucin 18	MUC18	T
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Mutated in colorectal cancers, MCC	MCC	G
MutL homolog 1	MLH1	G
MutS homolog 2	MSH2	G
MutS homolog 3	MSH3	G
Myelin protein peripheral 22	PMP22	S
Myelodysplasia syndrome 1 gene	MDS1	G
Myeloid leukemia factor-1	MLF1	I
N-acetyltransferase 1	NAT1	E
N-acetyltransferase 2	NAT2	E
NADPH-dependent cytochrome P450 reductase	POR	E
Nerve growth factor	NGF	G
Nerve growth factor receptor	NGFR	G
Neuregulin	HGL	G
Neurexin		N
Neurofibromin 1	NF1	G
Neurofibromin 2	NF2	G
Neurokinin A	NKNA	N
Neurokinin B	NKNB	N
Neuronal apoptosis inhibitory protein	NAIP	I
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Neurotensin	NTS	N
Neurotensin receptor	NTSR1	N
Neurotrophic tyrosine kinase receptor 1	NTRK1	G
Neutral endopeptidase		E
Niacin receptor		G
Nodal	NODAL	G
Norrie disease protein	NDP	G
Notch 3	NOTCH3	G



Notch ligand - jagged 1	JAG1, AGS	G
Nuclear factor kappa beta	NFKB	I
Nuclear factor of activated T cells (NFAT) complex, cytosolic	NFATC	G
Nuclear factor of activated T cells (NFAT) complex, preexisting component	NFATP	G
Nuclear mitotic apparatus protein 1	NUMA1	G
Nucleophosmin	NPM1	T
Oligophrenin-1	OPHN1	G
Oncogene abl1	ABL1	G
Oncogene abl2		G
Oncogene akt1		G
Oncogene akt2	AKT2	G
Oncogene axl	AXL	G
Oncogene bcl2		G
Oncogene bcr/abl		G
Oncogene B-lym		G
Oncogene B-raf		G
Oncogene clk1		G
Oncogene c-myc		G
Oncogene cot		G
Oncogene crk		G
Oncogene crkl		G
Oncogene ect2		G
Oncogene ELK1	ELK1	G
Oncogene ELK2	ELK2	G
Oncogene ems1		G
Oncogene ERB		G
Oncogene ERB2		G
Oncogene ERBA		G
Oncogene ERBAL2		G
Oncogene ERG (early reponse gene)		G
Oncogene ETS1		G
Oncogene ETS2		G
Oncogene EVI1	EVI1	G
Oncogene fes		G
Oncogene fgr		G
Oncogene fos	FOS	G
Oncogene fps		G
Oncogene GLI1	GLI	G
Oncogene GLI2	GLI2	G
Oncogene GLI3	GLI3	G
Oncogene gro1		G
Oncogene gro2		G
Oncogene Ha-ras	HRAS	G
Oncogene hs1		G
Oncogene hst	FGF4	G
Oncogene int1	WNT1	G

Oncogene int2	FGF3	G
Oncogene int3	Notch4	G
Oncogene int4	WNT3	G
Oncogene jun	JUN	G
Oncogene KIT	KIT, PBT	G
Oncogene LCO	LCO	G
Oncogene l-myc		G
Oncogene Ipsa		G
Oncogene lyn		G
Oncogene maf		G
Oncogene mas1		G
Oncogene mcf2		G
Oncogene mdm2	MDM2	G
Oncogene mel		G
Oncogene met	MET	G
Oncogene mos		G
Oncogene mpl		G
Oncogene MUM1	MUM1	G
Oncogene myb	MYB	G
Oncogene myc	MYC	G
Oncogene n-myc		G
Oncogene N-ras (neuroblastoma v-ras)	NRAS	G
Oncogene ovc		G
Oncogene pim1		G
Oncogene pti-1sea		G
Oncogene pvt1		G
Oncogene raf	RAF	G
Oncogene ralb		G
Oncogene rel		G
Oncogene ret	RET	G
Oncogene r-myc		G
Oncogene ros		G
Oncogene R-ras		G
Oncogene sis	PDGFB	G
Oncogene ski		G
Oncogene sno		G
Oncogene spi1		G
Oncogene src		G
Oncogene tc21		G
Oncogene TEL	ETV6	G
Oncogene tim		G
Oncogene vavtrk		G
Oncogene v-Ki-ras2	KRAS2	G
Oncogene yes		G
Oncogene yuasa		G
Oncostatin M	OSM	G
Oncostatin M receptor	OSMR	G
Opioid receptor, delta	OPRD1	N

Opioid receptor, kappa	OPRK1	N
Opioid receptor, mu	OPRM1	N
Orexin	OX	G
Osteopontin	OPN	G
Oxytocin	OXT	N
Oxytocin receptor	OXTR	N
Paired box homeotic gene 3	PAX3	G
Paired box homeotic gene 6	PAX6	G
Paired box homeotic gene 7	PAX7	G
Paired-like homeodomain transcription factor 2	PITX2	G
Paired-like homeodomain transcription factor 3	PITX3	G
Parathyroid hormone	PTH	G
Parathyroid hormone receptor	PTHr1	G
Parathyroid hormone related-peptide	PTHrP	G
Parvalbumin	PVALB	G
Patched (Drosophila) homolog, PTCH	PTCH	G
PCNA (proliferating cell nuclear antigen)		E
Peanut-like 1	PNUTL1	I
Peroxisome proliferative activated receptor, alpha	PPARA	T
Peroxisome proliferative activated receptor, gamma	PPARG	T
P-glycoprotein 1	PGY1	T
P-glycoprotein 3	PGY3	T
Phenylalanine hydroxylase	PAH	E
Phosphatase & tensin homolog	PTEN	G
Phosphatidylinositol glycan, class A (paroxysmal nocturnal hemoglobinuria)	PIGA	G
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C epsilon		I
Phosphomannomutase 1	PMM1	G
Phosphomannomutase 2	PMM2	G
Plasminogen	PLG	E
Plasminogen activator inhibitor 1	PAI1	E
Plasminogen activator inhibitor 2	PAI2	E
Plasminogen activator receptor, Urokinase	UPAR; PLAUR	S
Plasminogen activator, Tissue	PLAT; TPA	E
Plasminogen activator, Urokinase	UPA; PLAU	E
Platelet derived growth factor	PDGF	G
Platelet derived growth factor receptor	PDGFR	G
Platelet glycoprotein 1b, beta	GP1BB	I

Platelet glycoprotein 1b, gamma	GP1BG	I
Platelet glycoprotein IX	GP9	I
Platelet glycoprotein V	GP5	I
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium inwardly-rectifying channel J11	KCNJ11	N
Potassium voltage-gated channel A1	KCNA1	N
Potassium voltage-gated channel E1	KCNE1	N
Potassium voltage-gated channel Q1	KCNQ1	N
Potassium voltage-gated channel Q2	KCNQ2	N
Potassium voltage-gated channel Q3	KCNQ3	N
POU domain, class 1, transcription factor 1 (Pit1)	POU1F1	G
POU domain, class 3, transcription factor 4	POU3F4	G
POU domain, class 4, transcription factor 3	POU4F3	G
Pre-B-cell leukemia transcription factor 1	PBX1	G
Preproglucagon	GCG;GLP1; GLP2	G
Preproglucagon		T
Prion protein	PRNP	N
Prodynorphin		N
Progesterone receptor (RU486 binding receptor)	PGR	G
Prohibitin	PHB	G
Prolactin	PRL	G
Prolactin receptor	PRLR	G
Prolactin releasing hormone	PRH	G
Proliferin	PLF	G
Promyelocytic-leukemia gene	PML	G
Proopiomelanocortin	POMC	N
Prophet of Pit1	PROP1	G
Prostacyclin synthase		I
Prostaglandin 15-OH dehydrogenase	HGPD; PGDH	I
Prostaglandin D - DP receptor		I
Prostaglandin E1 receptor		I
Prostaglandin E2 receptor		I
Prostaglandin E3 receptor		I
Prostaglandin F - FP receptor		I
Prostaglandin IP receptor		I
Prostate cancer anti-metastasis gene KAI1	KAI1	G
Protein kinase B	PRKB	
Protein kinase C, alpha	PRKCA	E
Protein phosphatase 2, regulatory subunit A, beta isoform	PPP2R1B	E
Protein tyrosine phosphatase, non-receptor type 12	PTPN12	G
Purine nucleoside phosphorylase	NP	E
Purinergic receptor P1A1		N
Purinergic receptor P1A2		N
Purinergic receptor P1A3		N

Purinergic receptor P2X, 1	P2RX1	N
Purinergic receptor P2X, 2	P2RX2	N
Purinergic receptor P2X, 3	P2RX3	N
Purinergic receptor P2X, 4	P2RX4	N
Purinergic receptor P2X, 5	P2RX5	N
Purinergic receptor P2X, 6	P2RX6	N
Purinergic receptor P2X, 7	P2RX7	N
Purinergic receptor P2Y, 1	P2RY1	N
Purinergic receptor P2Y, 11	P2RY11	N
Purinergic receptor P2Y, 2	P2RY2	N
Rabphilin		N
RAD51, DNA repair protein	RAD51	G
RAD52, DNA repair protein	RAD52	G
RAD54, DNA repair protein	RAD54	G
RAD55, DNA repair protein	RAD55	G
RAD57, DNA repair protein	RAD57	G
RAS-associated protein, RAB3A	RAB3A	N
Ras-G-protein	RAS	G
Receptor tyrosine kinase (RTK), Nsk2	NSK2	G
Relaxin H1	RLN1	G
Relaxin H2	RLN2	G
Replication factor A		E
Replication factor C	RFC2	E
Retinoblastoma 1	RB1	G
Retinoic acid receptor, alpha	RARA	G
Retinoic acid receptor, beta	RARB	G
Retinoic acid receptor, gamma	RARG	G
Retinoschisis, X-linked, juvenile	RS	G
Rhabdoid tumors	SMARCB1	G
Ribonucleotide reductase, RRM		E
Ribosomal protein L13A	RPL13A	G
Ribosomal protein L17	RPL17	G
Ribosomal protein S6 kinase	RPS6KA3	E
RIGUI	RIGUI	G
Rim		N
Ryanodine receptor 1, skeletal	RYR1	G
S-adenosylmethionine decarboxylase, AMD		E
SAP (SLAM-associated protein)	SH2D1A	I
Secretin	SCT	T
Secretin receptor, SCTR	SCTR	T
Serine hydroxymethyltransferase	SHMT	E
Serine/threonine kinase 11	STK11	G
Serine/threonine kinase 2	STK2	G
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N

Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Signal transducer and activator of transcription 1	STAT1	G
Signal transducer and activator of transcription 2	STAT2	G
Signal transducer and activator of transcription 3	STAT3	G
Signal transducer and activator of transcription 4	STAT4	G
Signal transducer and activator of transcription 5	STAT5	G
Signaling lymphocyte activation molecule	SLAM	I
Sine oculis homeobox, drosophila, homolog 1	SIX1	G
Sine oculis homeobox, drosophila, homolog 2	SIX2	G
Sine oculis homeobox, drosophila, homolog 5	SIX5	G
Small nuclear ribonucleoprotein polypeptide N	SNRPN	S
Smoothed (Drosophila) homolog	SMOH	G
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage gated, type V, alpha polypeptide	SCN5A	N
Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Solute carrier family 1 (glutamate transporter), member 1	SLC1A1	T
Solute carrier family 1 (glutamate transporter), member 2	SLC1A2	T
Solute carrier family 12, member 1	SLC12A1	T
Solute carrier family 12, member 2	SLC12A2	T
Solute carrier family 12, member 3	SLC12A3	T
Solute carrier family 19 (folate transporter), member 1	SLC19A1	T
Solute carrier family 25, member 12	SLC25A12	T
Solute carrier family 5 (sodium/glucose transporter), member 1	SLC5A1	T
Solute carrier family 5 (sodium/glucose transporter), member 2	SLC5A2	T
Solute carrier family 5 (sodium/glucose transporter), member 5	SLC5A5	T

Solute carrier family 5, member 3	SLC5A3	T
Solute carrier family 6 (GAMMA-AMINOBTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Solute carrier family 6 (neurotransmitter transporter, serotonin), member 4	SLC6A4	T
Somatostatin	SST	N
Somatostatin receptor, SSTR1	SSTR1	N
Somatostatin receptor, SSTR2	SSTR2	G
Somatostatin receptor, SSTR3	SSTR3	N
Somatostatin receptor, SSTR4	SSTR4	N
Somatostatin receptor, SSTR5	SSTR5	N
Sorcin	SRI	T
SOS1 guanine nucleotide exchange factor	SOS1	G
SRY-box 11	SOX11	G
Stem cell factor	SCF	G
Steroid hormone receptor responsive DNA elements		G
Steroidogenic acute regulatory protein	STAR	T
Substance P		N
Sulfonylurea receptor	SUR	G
Suppression of tumorigenicity 3 gene	ST3	G
Suppression of tumorigenicity 8 gene	ST8	G
Surfeit 1	SURF1	G
Synapsin 1a & 1b	SYN1	N
Synapsin 2a & 2b	SYN2	N
Synaptic vesicle protein 2	SV2	N
Synaptobrevin 1	SYB1	N
Synaptobrevin 2	SYB2	N
Synaptogyrin		N
Synaptophysin	SYP	N
Synaptosomal-associated protein, 25KD	SNAP25	N
Synaptotagmin 1	SYT1	N
Synaptotagmin 2	SYT2	N
Syndecan 1	SYND1	G
Syndecan 2	SYND2	G
Syndecan 3	SYND3	G
Syndecan 4	SYND4	G
Synovial sarcoma gene 1	SSX1	G
Synovial sarcoma gene 2	SSX2	G
Syntaxin 1	STX1	N
Tachykinin receptor, NK1R	TACR1	N
Tachykinin receptor, NK2R	TACR2	N
Tachykinin receptor, NK3R	TACR3	N
Talin	TLN	G

Talin, TLN		S
T-cell acute lymphocytic leukemia 1	TAL1	I
T-cell acute lymphocytic leukemia 2	TAL2	I
T-cell receptor, alpha	TCRA	I
T-cell receptor, delta	TCRD	I
Telomerase protein component		E
Tenascin (cytotactin)		S
Tenascin XA	TNXA	S
Terminal deoxynucleotidyltransferase, TDT		E
Testis-specific protein Y	TSPY	G
Thrombopoietin	THPO	G
Thromboxane A synthase 1	TBXAS1	I
Thromboxane A2	TXA2	I
Thromboxane A2 receptor	TBXA2R	I
Thy-1 T-cell antigen	THY1	I
Thymidylate synthase	TYMS	E
Thymopoietin	TMPO	G
Thymosin		I
Thyroid-stimulating hormone receptor	TSHR	G
Thyroid-stimulating hormone, alpha	TSHA	G
Thyroid-stimulating hormone, beta	TSHB	G
Thyrotropin releasing hormone	TRH	N
Thyrotropin releasing hormone	TRH	G
Thyrotropin releasing hormone receptor	TRHR	N
Tip-associated protein	TAP	I
Tissue inhibitor of metalloproteinase 1, TIMP1	TIMP1	E
Tissue inhibitor of metalloproteinase 2, TIMP2	TIMP2	E
Tissue inhibitor of metalloproteinase 3, TIMP3	TIMP3	E
Tissue inhibitor of metalloproteinase 4, TIMP4	TIMP4	E
Topoisomerase II		E
Transacylase		E
Transcobalamin 1, TCN1		T
Transcobalamin 2, TCN2	TCN2	T
Transcription factor 1, hepatic	TCF1	G
Transcription factor 2, hepatic	TCF2	G
Transcription factor 3	TCF3	G
Transcription factor binding to IGHM enhancer 3	TFE3	G
Transcription termination factor, RNA polymerase 1	TTF1	G
Transcription termination factor, RNA polymerase 2	TTF2	G
Transcription termination factor, RNA polymerase 3	TTF3	G
Transferrin	TF	G
Transferrin receptor	TFRC	G
Transforming growth factor, alpha	TGFA	G
Transforming growth factor, beta 2	TGFB2	G



Transforming growth factor, beta induced	TGFB1	G
Transforming growth factor, beta receptor 2	TGFBR2	G
Translocation in renal carcinoma on chromosome 8 gene	TRC8	G
Tuberous sclerosis 1	TSC1	G
Tuberous sclerosis 2	TSC2	G
Tubulin		S
Tumor susceptibility gene 101	TSG101	G
Tumour necrosis factor (TNF) receptor associated factor 1	TRAF1	I
Tumour necrosis factor (TNF) receptor associated factor 2	TRAF2	I
Tumour necrosis factor (TNF) receptor associated factor 3	TRAF3	I
Tumour necrosis factor (TNF) receptor associated factor 4	TRAF4	I
Tumour necrosis factor (TNF) receptor associated factor 5	TRAF5	I
Tumour necrosis factor (TNF) receptor associated factor 6	TRAF6	I
Tumour necrosis factor alpha	TNFA	I
Tumour necrosis factor alpha receptor	TNFAR	I
Tumour necrosis factor beta	TNFB	I
Tumour necrosis factor beta receptor	TNFBR	I
Tumour protein p53	TP53, P53	G
Tumour protein p63	TP63	G
Tumour protein p73	TP73	G
Tumour protein, translationally-controlled 1	TPT1	G
Tumour suppressor gene DRA	DRA	I
Twist (Drosophila) homolog	TWIST	G
Ubiquitin		G
Ubiquitin activating enzyme, E1		E
Ubiquitin B	UBB	G
Ubiquitin C	UBC	G
Ubiquitin fusion degeneration 1-like	UFD1L	G
Ubiquitin protein ligase E3A	UBE3A	E
Vacuolar proton pump, subunit 1	VPP1	N
Vacuolar proton pump, subunit 3	VPP3	N
Vasoactive intestinal polypeptide	VIP	N
Vasoactive intestinal polypeptide receptor	VIPR	N
Vitamin B12-binding (R) protein		G
Vitamin D receptor	VDR	G
v-myc avian myelocytomatosis viral oncogene homolog	MYC	G
Von Hippel-Lindau gene	VHL	G
Werner syndrome helicase	WRN	G
Wilms tumour gene 1	WT1	G
Wilms tumour gene 2	WT2	G

Wilms tumour gene 4	WT4	G
Winged helix nude	WHN	G
Wiskott-Aldrich syndrome protein	WASP, THC	I
Xeroderma pigmentosum, complementation group B	XPB	E
Xeroderma pigmentosum, complementation group C	XPC	E
Xeroderma pigmentosum, complementation group D		E
Xeroderma pigmentosum, complementation group E		E
Xeroderma pigmentosum, complementation group F	XPF	E
Xeroderma pigmentosum, complementation group G	ERCC5	E
X-ray repair gene	XRCC9	G
YY1 transcription factor	YY1	G
Zinc finger protein 198	ZIC198	S
Zinc finger protein HRX	ALL1	I

36. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 35.
37. A set according to claim 35 or 36 in which a minority of said probes for listed genes are absent.
38. A set according to claim 35 or 36 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.
39. A set according to claim 35 or 36 in which a limited number of probes are replaced by probes for non-listed genes.
40. A set of probes for a core group of genes according to any of claims 35 to 39 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.
41. A set according to any of claims 35 to 40 consisting of probes for members of a sub-group of the core group.
42. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.

43. A set according to **any** preceding claim wherein said probes are on a substrate which forms **part of or** consists of one or more chip plate(s), for use in a chip assay for **detection of said gene variants**.
44. A set according to **any** preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.
45. A set according to claim 42 or 43 in which said substrate is a semiconductor microchip.
46. A set according to **any** preceding claim for use in a biological assay for detection of said gene variants.
47. A set according to **any** preceding claim for use in the measurement of differential gene expression levels.
48. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
49. A medical device including a set according to any of claims 35 to 47 for use in an array for detection of differential gene expression levels.
50. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 35) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 35 and 37 to 47 and relating the probe hybridisation pattern to said variations.
51. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 36) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 36 to 47 and relating the probe interaction pattern to said variations.
52. Use of a set or device according to any of claims 35 to 47 for the prognosis and management of patients suffering from or at risk of developing symptoms and consequences of cancer.
53. Use of a set or device according to any of claims 35 to 47 for predicting likely therapeutic response and adverse events following therapeutic intervention.
54. Use of a set or device according to any of claims 35 to 47 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
55. Use of a set or device according to any of claims 35 to 47 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
56. Use of a set or device according to any of claims 35 to 47 for general health screening, occupational health purposes, healthcare planning on a population basis and other **healthcare** management utilisations.
57. Use of a set or **device** according to any of claims 35 to 47 for the development of new strategies of **therapeutic** intervention and in clinical trials.
58. Use of a set or device according to any of claims 35 to 47 for construction of and generation of algorithms for patient and healthcare management.
59. Use of a set or device according to any of claims 35 to 47 for modelling or assessing the impact of diseases or healthcare management strategies on

- individuals, groups, patient cohorts or populations
60. Use of a set or device according to any of claims 35 to 47 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
  61. Use of a set or device according to any of claims 35 to 47 for predicting optimum configuration/management of thereapeutic intervention.
  62. A method according to claim 50 or 51 in which the identification of gene variants is indicative of a higher risk of developing symptoms and consequences of cancer for the patient or individual.
  63. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop symptoms and consequences of cancer which method comprises:
    - i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from cancer;
    - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the cancer;
    - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 35 to 41;
    - iv) calculating the frequencies of these alleles in the samples from i) and ii);
    - v) comparing the frequencies of these alleles in i) and ii);
    - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing symptoms and consequences of cancer.
  64. A method for assessing whether a given subject will be at risk of developing symptoms and consequences of cancer, which comprises comparing said subject's genotype with a model generated by the method of claim 63.
  65. A method according to any of claims 50, 51, 63 and 64 wherein at least one step is computer-controlled.
  66. An assay suitable for use in a method according to any of claims 50, 51, 63 and 64; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 35 to 41 in a biological sample.
  67. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing symptoms and consequences of cancer; said kit comprising:
    - i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 35 or 37 to 41 in a sample of human DNA;
    - ii) reagents for use in the detection process
    - iii) readout indicating the probability of a patient or individual developing symptoms and consequences of cancer.
  68. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing symptoms and consequences of cancer; said kit comprising:
    - i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core

- group of genes as defined in any of claims 36 to 41 in an expressed-protein-containing human sample;
- ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing symptoms and consequences of cancer.
69. A set of probes according to claim 35, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.
70. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to CNS dysfunction, damage or disease; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

#### KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME  
 T TRANSPORT & STORAGE  
 S STRUCTURAL  
 I IMMUNITY  
 N NERVOUS TRANSMISSION  
 G GROWTH & DIFFERENTIATION

CNS GENE LIST	HUGO gene symbol	Protein function
11beta hydroxysteroid dehydrogenase 2	HSD11B2	E
2,3-bisphosphoglycerate mutase	BPGM	E
2,4-dienoyl CoA reductase	DECR	E
3 beta hydroxysteroid dehydrogenase 2	HSD3B2	E
3-oxoacid CoA transferase	OXCT	E
4-hydroxyphenylpyruvate dioxygenase	HPD	E
5,10-methylenetetrahydrofolate reductase (NADPH)	MTHFR	E
6-pyruvoyltetrahydropterin synthase	PTS	E
Acetoacetyl 2-CoA-thiolase	ACAT2	E
Acetyl CoA acyltransferase	ACAA	E
Acetyl CoA carboxylase alpha	ACACA	E
Acetylcholine receptor, nicotinic, alpha A1	CHRNA1	N
Acetylcholine receptor, nicotinic, alpha A2	CHRNA2	N
Acetylcholine receptor, nicotinic, alpha A3	CHRNA3	N
Acetylcholine receptor, nicotinic, alpha A4	CHRNA4	N
Acetylcholine receptor, nicotinic, alpha A5	CHRNA5	N
Acetylcholine receptor, nicotinic, alpha A6	CHRNA6	N
Acetylcholine receptor, nicotinic, alpha A7	CHRNA7	N
Acetylcholine receptor, nicotinic, beta 1	CHRNA1	N
Acetylcholine receptor, nicotinic, beta 2	CHRNA2	N

Acetylcholine receptor, nicotinic, beta 3	CHRNA3	N
Acetylcholine receptor, nicotinic, beta 4	CHRNA4	N
Acetylcholine receptor, nicotinic, epsilon	CHRNAE	N
Acetylcholine receptor, nicotinic, gamma	CHRNAG	N
Acetylcholinesterase	ACHE	E
Acyl CoA dehydrogenase, long chain	ACADL	E
Acyl CoA dehydrogenase, medium chain	ACADM	E
Acyl CoA dehydrogenase, short chain	ACADS	E
Acyl-CoA thioesterase		E
Adaptin, beta 3A	ADTB3A	T
Adducin, alpha	ADD1	S
Adducin, beta	ADD2	S
Adenosine monophosphate deaminase	AMPD	E
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N
Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenyl cyclase		N
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adenylosuccinate lyase	ADSL	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
Adrenocorticotrophic hormone (ACTH) receptor	ACTHR	G
Adrenoleukodystrophy gene	ALD	E
Albumin, ALB	ALB	T
Aldehyde dehydrogenase 10	ALDH10	E
Aldolase A	ALDOA	E
Aldolase B	ALDOB	E
Aldolase C	ALDOC	E
Aldosterone receptor	MLR	G
Alpha 2 macroglobulin	A2M	I
alpha tectorin	TECTA	G
alpha thalassemia gene	ATRX	N
alpha1-antitrypsin	PI	E
alpha2-antiplasmin	PLI	E
alpha-Galactosidase A	GLA	E

alpha-ketoglutarate dehydrogenase		E
alpha-L-Iduronidase	IDUA	E
alpha-synuclein	SNCA	N
Aminomethyltransferase	AMT	E
Aminopeptidase P	XPNPEP2	E
Amylo-1,6-glucosidase	AGL	E
Amyloid beta (A4) precursor protein-binding, APBB1	APBB1	N
Amyloid beta A4 precursor protein	APP	N
Amyloid beta A4 precursor-like protein	APLP	N
Angiopoietin 1	ANGPT1	G
Angiopoietin 2	ANGPT2	G
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensin receptor 1	AGTR1	T
Angiotensin receptor 2	AGTR2	T
Angiotensinogen	AGT	E
Antidiuretic hormone receptor	ADHR	T
Antithrombin III	AT3	E
Apolipoprotein A I	APOA1	T
Apolipoprotein A II	APOA2	T
Apolipoprotein B	APOB	T
Apolipoprotein C1	APOC1	T
Apolipoprotein C2	APOC2	T
Apolipoprotein C3	APOC3	T
Apolipoprotein D	APOD	T
Apolipoprotein E	APOE	T
Apolipoprotein H	APOH	T
Archaete-scute homolog 2	ASH2	G
Arginase	ARG1	E
Arginine vasopressin	AVP	N
Arginosuccinate lyase	ASL	E
Arginosuccinate synthetase	ASS	E
Arylsulfatase A	ARSA	E
Arylsulfatase B	ARSB	E
Arylsulfatase D	ARSD	E
Arylsulfatase E	ARSE	E
Arylsulfatase F	ARSF	E
Aspartoacylase	ASPA	E
Aspartylglucosaminidase	AGA	E
Astrotactin	ASTN	G
Ataxia telangiectasia complementation group D	ATD, ATDC	G
Ataxia telangiectasia gene, AT	ATM	G
ATP-binding cassette transporter 7	ABC7	I
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G
Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G

Bagpipe homeobox, <i>drosophila</i> homolog of, 1	BAPX1	G
beta-Glucuronidase	GUSB	E
beta-synuclein	SNCB	N
Bilirubin UDP-glucuronosyltransferase		E
Bloom syndrome protein	BLM	G
Bradykinin receptor B1		I
Bradykinin receptor B2		I
Brain derived neurotrophic factor	BDNF	G
Brain derived neurotrophic factor (BDNF) receptor	BDNFR	G
Butyrylcholinesterase	BCHE	E
Ca(2+) transporting ATPase, slow twitch	ATP2A2	T
Cadherin E	CDH1	G
Cadherin EP		G
Cadherin N	CDH2	G
Cadherin P	CDH3	G
Calbindin 1	CALB1	G
Calbindin D9K	CALB3	G
Calcineurin A1	CALNA1	I
Calcineurin A2	CALNA2	I
Calcineurin A3	CALNA3	I
Calcineurin B		I
Calcitonin/Calcitonin gene-related peptide alpha	CALCA	N
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha-1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha-1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha-1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha-1E (CACNL1A6)	CACNA1E	N
Calcium channel, voltage-dependent, Alpha-2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, L type, alpha 1S subunit	CACNA1S	N
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N
Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	CACNA1A	N
Calcium channel, voltage-dependent, T-type		N
Calmodulin 1	CALM1	G
Calmodulin 2	CALM2	G
Calmodulin 3	CALM3	G



Calmodulin-dependant protein kinase II	CAMK2A	G
Calnexin	CANX	G
Calpain	CAPN, CAPN3	E
Calretinin	CALB2	N
Cannabinoid receptor	CNR1	N
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Cardiac-specific homeobox, CSX	CSX	G
Carnitine acetyltransferase	CRAT	E
Carnitine acylcarnitine translocase	CACT	E
Carnitine transporter protein	CDSP, SCD	T
Carnosinase		N
Caspase 1	CASP1	G
Catechol-O-methyltransferase	COMT	E
CD1	CD1	I
CD4	CD4	I
Cell adhesion molecule, neural, NCAM1	NCAM1	G
Cell adhesion molecule, neural, NCAM120	NCAM120	G
Cell adhesion molecule, neural, NCAM2	NCAM2	G
Ceroid lipofuscinosis neuronal 2	CLN2	N
Ceroid lipofuscinosis neuronal 3	CLN3	N
Ceroid lipofuscinosis neuronal 4	CLN4	N
Ceroid lipofuscinosis neuronal 5	CLN5	N
Ceroid lipofuscinosis neuronal 6	CLN6	N
Chemokine receptor CCR2	CCR2	I
Chemokine receptor CCR3	CCR3	I
Chemokine receptor CCR5	CCR5	I
Chemokine receptor CXCR4	CXCR4	I
Chloride channel 1, skeletal muscle	CLCN1	S
Cholecystokinin	CCK	N
Cholecystokinin B receptor	CCKBR	N
Choline acetyltransferase	CHAT	E
Choroideremia gene	CHM	S
Chromogranin A	CHGA	G
Chymotrypsinogen		E
Ciliary neurotrophic factor (CNTF)	CNTF	G
Ciliary neurotrophic factor (CNTF) receptor	CNTR	G
Clathrin		T
CoA transferase		E
Cochlin	COCH	I
Cockayne syndrome gene, CKN1	CKN1	G
Cofilin		S
Collagen I alpha 1	COL1A1	S
Collagen I alpha 2	COL1A2	S
Collagen II alpha 1	COL2A1	S
Collagen III alpha 1	COL3A1	S

Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E
Cyclic nucleotide phosphodiesterase 8	PDE8	E
Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclin-dependent kinase 2	CDK2	G
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
CYP11A1	CYP11A1	E
CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E
CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E
CYP2C18	CYP2C18	E
CYP2C19	CYP2C19	E
CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E
CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E
CYP2F1	CYP2F1	E
CYP2J2	CYP2J2	E
CYP3A3	CYP3A3	E
CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystathionase	CTH	E
Cystathione beta synthase	CBS	E
Cystatin B	CSTB	T

Electron-transferring-flavoprotein alpha	ETFA	T
Electron-transferring-flavoprotein beta	ETFB	T
Electron-transferring flavoprotein dehydrogenase	ETFDH	E
Emerin	EMD	T
Empty spiracles (drosophila) homologue 1	EMX1	G
Empty spiracles (drosophila) homologue 2	EMX2	G
Endobrevin	VAMP8	N
Endothelin 1	EDN1	N
Endothelin 2	EDN2	N
Endothelin 3	EDN3	N
Endothelin converting enzyme	ECE1	N
Endothelin receptor type A	EDNRA	N
Endothelin receptor type B	EDNRB	N
Enolase	ENO1	E
Enoyl CoA isomerase		E
Enoyl CoA reductase		E
Enterokinase	PRSS7, ENTK	E
Ephrin-A	EFNA	G
Ephrin-B	EFNB	G
Epidermal growth factor	EGF	G
Epidermal growth factor receptor	EGFR	G
Epilepsy, progressive myoclonic 2 gene	EPM2A	E
EWS RNA-binding protein	EWSR1	G
Excision repair complementation group 4 protein	ERCC4	E
Exostosin 1	EXT1	S
Exostosin 2	EXT2	S
Factor 1 (No. one)	F1	I
Factor III	F3	I
Factor IX	F9	I
Factor V	F5	I
Factor VII	F7	I
Factor VIII	F8	I
Factor X	F10	I
Factor XI	F11	I
Factor XII	F12	I
Factor XIII A & B	F13A & F13B	I
Fanconi anemia, complementation group A	FANCA	T
Fanconi anemia, complementation group C	FANCC	T
Fanconi anemia, complementation group D	FANCD	T
Fibrillin 2	FBN2	G
Fibrinogen alpha	FGA	S
Fibrinogen beta	FGB	S
Fibrinogen gamma	FGG	S
Fibroblast growth factor	FGF1	G
Fibroblast growth factor receptor 1	FGFR1	G
Fibroblast growth factor receptor 2	FGFR2	G

Fibroblast growth factor receptor 3	FGFR3	G
Fibronectin precursor	FN1	G
Flightless-II, Drosophila homolog of	FLII	G
Follicle stimulating hormone receptor	FSHR, ODG1	G
Follicle stimulating hormone, FSH	FSHB	G
Forkhead transcription factor 10	FKHL10	G
Formiminotransferase		E
Fragile site, folic acid type, rare, fra(X) A	FRAXA	N
Fragile site, folic acid type, rare, fra(X) E	FRAXE	N
Fragile site, folic acid type, rare, fra(X) F	FRAXF	N
Frataxin	FRDA	G
Fructose-1,6-diphosphatase	FBP1	E
Fukuyama type congenital muscular dystrophy	FCMD	G
GABA receptor, alpha 1	GABRA1	N
GABA receptor, alpha 2	GABRA2	N
GABA receptor, alpha 3	GABRA3	N
GABA receptor, alpha 4	GABRA4	N
GABA receptor, alpha 5	GABRA5	N
GABA receptor, alpha 6	GABRA6	N
GABA receptor, beta 1	GABRB1	N
GABA receptor, beta 2	GABRB2	N
GABA receptor, beta 3	GABRB3	N
GABA receptor, gamma 1	GABRG1	N
GABA receptor, gamma 2	GABRG2	N
GABA receptor, gamma 3	GABRG3	N
GABA transaminase	ABAT	E
Galactocerebrosidase	GALC	E
Galactose 1-phosphate uridyl-transferase	GALT	E
Galactosyltransferase 1	GT1	G
Galactosyltransferase, alpha 1,3	GGTA1	G
Galactosyltransferase, beta 3	B3GALT	G
Galanin	GAL	N
Galanin receptor	GALNR1	N
Gamma-glutamyltransferase 1	GGT1	T
Gap junction protein beta 2	GJB2	T
Gap junction protein beta 3	GJB3	T
Gastric Intrinsic factor, GIF	GIF	E
Gastrulation brain homeobox 2	GBX2	G
Geniospasm 1	GSM1	G
Gephyrin		N
Glial-cell derived neurotrophic factor (GDNF) receptor		N
Glial-cell derived neurotrophic factor, GDNF	GDNF	N
Glucosidase, acid alpha	GAA	E
Glutamate decarboxylase, GAD	GAD1	E
Glutamate dehydrogenase	GLUD1	E
Glutamate receptor 1	GLUR1	N

Glutamate receptor 2	GLUR2	N
Glutamate receptor 3	GLUR3	N
Glutamate receptor 4	GLUR4	N
Glutamate receptor 5	GLUR5	N
Glutamate receptor 6	GLUR6	N
Glutamate receptor 7	GLUR7	N
Glutamate receptor, ionotropic, NMDA 1	NMDAR1	N
Glutamate receptor, ionotropic, NMDA 2A	NMDAR2A	N
Glutamate receptor, ionotropic, NMDA 2B	NMDAR2B	N
Glutamate receptor, ionotropic, NMDA 2C	NMDAR2C	N
Glutamate receptor, ionotropic, NMDA 2D	NMDAR2D	N
Glutamate-cysteine ligase	GLCLC	E
Glutaryl-CoA dehydrogenase	GCDH	E
Glutathione	GSH	T
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glutathione synthetase	GSS	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
Glycerol kinase	GK	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
Glycine dehydrogenase	GLDC	E
Glycine receptor, alpha	GLRA2	N
Glycine receptor, beta		N
Glycine transporter	GLYT	N
Glycogen phosphorylase	PYGL	E
GM2 ganglioside activator protein, GM2A	GM2A	E
Gonadotropin releasing hormone receptor	GNRHR	G
GTP cylcohydrolase 1	GCH1	G
Guanidinoacetate N-methyltransferase	GAMT	E
Guanine nucleotide-binding protein, alpha activating activity polypeptide, GNAO	GNAO1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 1, GNAI1	GNAI1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 2, GNAI2	GNAI2	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 3, GNAI3	GNAI3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS1	GNAS1	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS2	GNAS2	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS3	GNAS3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS4	GNAS4	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT1	GNAT1	N

Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT2	GNAT2	N
Guanine nucleotide-binding protein, beta polypeptide 3	GNB3	N
Guanine nucleotide-binding protein, gamma polypeptide 5	GNG5	N
Guanine nucleotide-binding protein, q polypeptide	GNAQ	N
Guanylate cyclase 2D, membrane (retina-specific)	GUCY2D	E
Guanylate cyclase activator 1A (retina)	GUCA1A	E
Guanylate kinase		E
Guanylyl cyclase		E
Gustducin, alpha (taste-specific G protein)	GDCA	N
Haeme regulated inhibitor kinase		E
Haemoglobin alpha 1	HBA1	T
Haemoglobin alpha 2	HBA2	T
Haemoglobin beta	HBB	T
Haemoglobin delta	HBD	T
Haemoglobin gamma A	HBG1	T
Haemoglobin gamma B	HBG2	T
Haemoglobin gamma G	HBGG	T
Heat shock protein, HSP60		I
Heat shock protein, HSP70		I
Heat shock protein, HSP90		I
Heat shock protein, HSPA1		I
Heat shock protein, HSPA2		I
Heparan sulfamidase		E
Heparin binding epidermal growth factor	HBEGF	G
Heparin Cofactor II	HCF2	I
Hepatic lipase	LIPC	E
Hexosaminidase A	HEXA,TSD	E
Hexosaminidase B	HEXB	E
Hippocampal cholinergic neurostimulating peptide, HCNP		N
Histamine receptors, H1		N
Histamine receptors, H2		N
Histamine receptors, H3		N
Histidase		E
HLA-B associated transcript 1	BAT1	I
HLH transcription factor HAND1	HAND1	G
HLH transcription factor HAND2	HAND2	G
HMG-CoA lyase	HMGCL	E
HMG-CoA reductase	HMGCR	E
Holocarboxylase synthetase	HLCS	E
Homeobox HB9	HLXB9	G
Human atonal gene	ATOH1	G
Hypoxanthine-guanine phosphoribosyltransferase, HGPRT	HPRT	E

Hypoxia inducible factor 1	HIF1A	E
Hypoxia inducible factor 2		E
IC7 A and B		I
Inositol 1,4,5-triphosphate receptor 1	ITPR1	G
Inositol monophosphatase	IMPA1	N
Inositol polyphosphate 1-phosphatase	INPP1	N
Insulin	INS	G
Insulin receptor	INSR	G
Insulin-like growth factor 1	IGF1	G
Insulin-like growth factor 1 receptor	IGF1R	G
Insulin-like growth factor 2	IGF2	G
Insulin-like growth factor 2 receptor	IGF2R	G
Integrin beta 1	ITGB1	G
Integrin beta 2	ITGB2	G
Integrin beta 3	ITGB3	G
Integrin, alpha 1	ITGA1	G
Integrin, alpha M	ITGAM	G
Inter-alpha-trypsin inhibitor, IATI		E
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) 10	IL10	I
Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I
Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
IP3 kinase		E

Isovaleric acid CoA dehydrogenase	IVD	E
Kallikrein 3	KAK3	I
Kallman syndrome gene 1	KAL1	G
Ketohexokinase	KHK	E
Kininogen, High molecular weight	KNG	I
Kynureninase		E
L1 cell adhesion molecule	L1CAM	N
Laminin 5, alpha 3	LAMA3	G
Laminin 5, beta 3	LAMB3	G
Laminin 5, gamma 2	LAMC2	G
Laminin M	LAMM	G
Laminin receptor 1	LAMR1	G
Latent transforming growth factor-beta binding protein 2	LTBP2	G
Leptin	LEP	G
Leptin receptor	LEPR	G
Leukaemia inhibitory factor	LIF	G
Leukaemia inhibitory factor receptor	LIFR	G
Leukin		I
Leukocyte-specific transcript 1	LST-1	I
Leukotriene A4 hydrolase		I
Leukotriene A4 synthase	LTA4S	E
Leukotriene B4 receptor		I
Leukotriene B4 synthase	LTB4S	E
Leukotriene C4 receptor		I
Leukotriene C4 synthase	LTC4S	E
Leukotriene D4/E4 receptor		I
LIM homeobox protein 1	LHX1	G
LIM homeobox protein 2	LHX2	G
LIM homeobox protein 3	LHX3	G
LIM homeobox protein 4	LHX4	G
Limbic associated membrane protein	LAMP	G
LIM-domain only protein 1	LMO1	G
LIM-domain only protein 2	LMO2	G
LIM-domain only protein 3	LMO3	G
LIM-domain only protein 4	LMO4	G
LIM-Kinase I (LINK-I)		I
Lipoprotein receptor, Low Density	LDLR	T
Lipoprotein, High Density	HDLDT1	T
Lipoprotein, Intermediate Density		T
Lipoprotein, Low Density 1		T
Lipoprotein, Low Density 2		T
Lipoprotein, Very Low Density	VLDLR	T
Low density lipoprotein receptor-related protein precursor	LRP	T
Lymphoid enhancer-binding factor	LEF-1	G
MAD (mothers against decapentaplegic, Drosophila) homologue 4	MADH4	G



Malonyl CoA decarboxylase		E
Mannosidase, alpha B lysosomal	MANB	E
Mannosidase, beta A lysosomal	MANBA	E
Marenostrin	MEFV	T
Melatonin receptor 1A	MTNR1A	N
Melatonin receptor 1B	MTNR1B	N
Methylguanine-DNA methyltransferase	MGMT	E
Methylmalonyl-CoA mutase	MUT	E
Mevalonate kinase	MVK	E
Microsomal triglyceride transfer protein	MTP	T
Microtubule associated protein	MAP	S
Mismatch repair gene, PMSL2	PMS2	G
Molybdenum cofactor synthesis 1	MOCS1	E
Molybdenum cofactor synthesis 2	MOCS2	E
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Msh homeobox homolog 2	MSX2	G
Mucopolidoses	GNPTA	E
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Myelin protein peripheral 22	PMP22	S
Myelin protein zero	MPZ	S
Myogenic factor 3	MYF3	G
Myogenic factor 4	MYF4	G
Myogenic factor 5	MYF5	G
Myosin 15	MYO15	S
Myosin 6	MYO6	S
Myosin 7A	MYO7A	S
Myotubularin	MTM1	S
Na <sup>+</sup> , K <sup>+</sup> ATPase, alpha	ATP1A1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 1	ATP1B1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 2	ATP1B2	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 3	ATP1B3	G
N-acetylglucosamine-6-sulfatase	GNS	E
N-acetylglucosaminidase, alpha	NAGLU	E
NADH dehydrogenase		E
NADPH-dependent cytochrome P450 reductase	POR	E
NB6		I
Nebulin	NEB	S
Necdin	NDN	G
Nerve growth factor	NGF	G
Nerve growth factor receptor	NGFR	G
Neural retina-specific gene	NRL	G
Neuraminidase sialidase	NEU	T

Neuregulin	HGL	G
Neurite growth-promoting factor 2	MDK	N
Neurite inhibitory protein		N
Neuroendocrine convertase 1	NEC1, PCSK1	E
Neurofibromin 1	NF1	G
Neurofibromin 2	NF2	G
Neurofilament protein, heavy	NFH	S
Neurofilament protein, NF125	NF150	S
Neurofilament protein, NF200	NF200	S
Neurofilament protein, NF68	NF68	S
Neurokinin A	NKNA	N
Neurokinin B	NKNB	N
Neuronal apoptosis inhibitory protein	NAIP	I
Neuronal molecule-1		I
Neuronal molecule-1 receptor		I
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Neurotensin	NTS	N
Neurotensin receptor	NTSR1	N
Neutral endopeptidase		E
Niemann-Pick disease protein	NPC1	T
Nitric oxide synthase 1, NOS1	NOS1	E
Nitric oxide synthase 2, NOS2	NOS2	E
Nitric oxide synthase 3, NOS3	NOS3	E
Notch 1	NOTCH1	G
Notch 2	NOTCH2	G
Notch 3	NOTCH3	G
Notch ligand - jagged 1	JAG1, AGS	G
Nuclear factor I-kappa-B-like gene	IKBL	I
Nucleoside diphosphate kinase-A	NDPKA	E
Oncogene bcl2		G
Oncogene GLI1	GLI	G
Oncogene GLI2	GLI2	G
Oncogene GLI3	GLI3	G
Oncogene sis	PDGFB	G
Opioid receptor, delta	OPRD1	N
Opioid receptor, kappa	OPRK1	N
Opioid receptor, mu	OPRM1	N
Ornithine delta-aminotransferase	OAT	E
Ornithine transcarbamoylase	OTC, NME1	E
Orthodenticle (Drosophila) homolog 1	OTX1	G
Orthodenticle (Drosophila) homolog 2	OTX2	G
Otoferlin	OTOF	N
Paired box homeotic gene 2	PAX2	G
Paired box homeotic gene 3	PAX3	G
Palmitoyl-protein thioesterase	PPT	T
Parkin	PARK2	N

Patched (Drosophila) homolog, PTCH	PTCH	G
Peanut-like 1	PNUTL1	I
Peptidylglycine alpha-amidating monooxygenase	PAM	E
Peripherin, PRPH		S
Peroxisomal membrane protein 1	PXMP1	S
Peroxisomal membrane protein 3	PXMP3	T
Peroxisome biogenesis factor 1	PEX1	T
Peroxisome biogenesis factor 19	PEX19	T
Peroxisome biogenesis factor 6	PEX6	T
Peroxisome biogenesis factor 7	PEX7	T
Peroxisome receptor 1	PXR1	T
Persyn		S
Phosphate regulating gene with homologies to endopeptidases on the X chromosome	PHEX	G
Phosphatidylinositol transfer protein	PITPN	G
Phosphoglucose isomerase	GPI	E
Phosphoglycerate kinase 1	PGK1	E
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C alpha		I
Phospholipase C beta		I
Phospholipase C delta	PLCD1	I
Phospholipase C epsilon		I
Phospholipase C gamma	PLCG1	I
Phosphomannomutase 2	PMM2	G
Phosphoribosyl pyrophosphate synthetase	PRPS1	E
Phytanoyl-CoA hydroxylase	PHYH	G
Plakophilin 1	PKP1	T
Plasminogen	PLG	E
Plasminogen activator inhibitor 1	PAI1	E
Plasminogen activator inhibitor 2	PAI2	E
Plasminogen activator receptor, Urokinase	UPAR; PLAUR	S
Plasminogen activator, Tissue	PLAT; TPA	E
Plasminogen activator, Urokinase	UPA; PLAU	E
Platelet derived growth factor	PDGF	G
Platelet derived growth factor receptor	PDGFR	G
Platelet-activating factor receptor	PAFR	I
Plectin 1	PLEC1	T
Postsynaptic density-95 protein	PSD95	N
Potassium channel, calcium-activated,	KCNN4	N
Potassium channel, subfamily K, member 1	KCNK1	N

Potassium channel, subfamily K, member 2	KCNK2	N
Potassium channel, subfamily K, member 3	KCNK3	N
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium voltage-gated channel A1	KCNA1	N
Potassium voltage-gated channel E1	KCNE1	N
Potassium voltage-gated channel Q1	KCNQ1	N
Potassium voltage-gated channel Q2	KCNQ2	N
Potassium voltage-gated channel Q3	KCNQ3	N
Potassium voltage-gated channel Q4	KCNQ4	N
POU domain, class 1, transcription factor 1 (Pit1)	POU1F1	G
POU domain, class 3, transcription factor 4	POU3F4	G
POU domain, class 4, transcription factor 3	POU4F3	G
Prekallikrein		I
Preproenkephalin	PENK	N
Presenilin 1	PSEN1	T
Presenilin 2	PSEN2	T
Prion protein	PRNP	N
Procollagen N-protease		E
Proline dehydrogenase	PRODH	E
Pro-melanin-concentrating hormone	PMCH	G
Proopiomelanocortin	POMC	N
Prosaposin	PSAP	N
Prostacyclin synthase		I
Prostaglandin 15-OH dehydrogenase	HGPD; PGDH	I
Prostaglandin D - DP receptor		I
Prostaglandin E1 receptor		I
Prostaglandin E2 receptor		I
Prostaglandin E3 receptor		I
Prostaglandin F - FP receptor		I
Prostaglandin I2 receptor		T
Prostaglandin IP receptor		I
Protease nexin 2	PN2	E
Protective protein for beta-galactosidase	PPGB	E
Protein C	PROC	I
Protein C inhibitor	PCI	I
Protein kinase C, alpha	PRKCA	E
Protein kinase C, gamma	PRKCG	E
Protein kinase G		E
Protein phosphatase 1, regulatory (inhibitor) subunit 3	PPP1R3	E
Protein S	PROS1	I
Prothrombin precursor	F2	I
Purine nucleoside phosphorylase	NP	E
Pyrroline-5-carboxylate synthetase	PYCS	E
Pyruvate carboxylase	PC	E
Pyruvate decarboxylase	PDHA	E
Ras-G-protein	RAS	G

Rathke pouch homeobox, RPX	RPX	G
Renin	REN	E
Replication factor C	RFC2	E
Retinal pigment epithelium specific protein (65kD)	RPE65	S
Retinaldehyde binding protein 1	RLBP1	T
Retinoblastoma 1	RB1	G
Rhodopsin kinase	RHOK	E
RIGUI	RIGUI	G
S100 calcium-binding protein A1	S100A1	N
S100 calcium-binding protein A2	S100A2	N
S100 calcium-binding protein A3	S100A3	N
S100 calcium-binding protein A4	S100A4	N
S100 calcium-binding protein A5	S100A5	N
S100 calcium-binding protein A6	S100A6	N
S100 calcium-binding protein A7	S100A7	N
S100 calcium-binding protein A8	S100A8	N
S100 calcium-binding protein A9	S100A9	N
S100 calcium-binding protein B	S100B	N
S100 calcium-binding protein P	S100P	N
Secretase, alpha		N
Secretase, beta		N
Secretase, gamma		N
Selectin E	SELE	N
Selectin L	SELL	N
Selectin P	SELP	N
Semaphorin A4	SEMA4	S
Semaphorin A5	SEMA5	S
Semaphorin D		S
Semaphorin E	SEMAE	S
Semaphorin F	SEMA3/F	S
Semaphorin W	SEMAW	S
Serotonin N-acetyltransferase	SNAT	E
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Signaling lymphocyte activation molecule	SLAM	I

Slug protein		G
Small nuclear ribonucleoprotein polypeptide N	SNRPN	S
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage gated, type IV, alpha polypeptide	SCN4A	N
Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Solute carrier family 1 (amino acid transporter), member 6	SLC1A6	T
Solute carrier family 1 (glial high affinity glutamate transporter), member 3	SLC1A3	T
Solute carrier family 1 (glutamate transporter), member 1	SLC1A1	T
Solute carrier family 1 (glutamate transporter), member 2	SLC1A2	T
Solute carrier family 12, member 1	SLC12A1	T
Solute carrier family 12, member 2	SLC12A2	T
Solute carrier family 12, member 3	SLC12A3	T
Solute carrier family 16 (monocarboxylate transporter), member 1	SLC16A1	T
Solute carrier family 16 (monocarboxylate transporter), member 7	SLC16A7	T
Solute carrier family 18, member 3	SLC18A3	T
Solute carrier family 2 (facilitated glucose transporter), member 1	SLC2A1	T
Solute carrier family 20, member 3	SLC20A3	T
Solute carrier family 25, member 12	SLC25A12	T
Solute carrier family 4 (anion exchanger), member 1	SLC4A1	T
Solute carrier family 4 (anion exchanger), member 2	SLC4A2	T
Solute carrier family 4 (anion exchanger), member 3	SLC4A3	T
Solute carrier family 5 (sodium/glucose transporter), member 1	SLC5A1	T
Solute carrier family 5 (sodium/glucose transporter), member 2	SLC5A2	T
Solute carrier family 5 (sodium/glucose transporter), member 5	SLC5A5	T
Solute carrier family 5, member 3	SLC5A3	T
Solute carrier family 6 (GAMMA-AMINO BUTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter	SLC6A3	T

transporter, dopamine), member 3		
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Solute carrier family 6 (neurotransmitter transporter, serotonin), member 4	SLC6A4	T
Solute carrier family 6, member 6	SLC6A6	T
Solute carrier family 7(amino acid transporter), member 1	SLC7A1	T
Solute carrier family 7(amino acid transporter), member 2	SLC7A2	T
Solute carrier family 7(amino acid transporter), member 7	SLC7A7	T
Somatostatin	SST	N
Somatostatin receptor, SSTR1	SSTR1	N
Somatostatin receptor, SSTR2	SSTR2	G
Somatostatin receptor, SSTR3	SSTR3	N
Somatostatin receptor, SSTR4	SSTR4	N
Somatostatin receptor, SSTR5	SSTR5	N
Spastic paraplegia 7	SPG7	G
Spectrin beta	SPTB	S
Sphingomyelinase	SMPD1	E
Spinocerebellar ataxia 8 gene	SCA8	N
SRY-box 11	SOX11	G
Steroid 5 alpha reductase 1	SRD5A1	E
Steroid 5 alpha reductase 2	SRD5A2	E
Steroid sulphatase	STS	E
Substance P		N
Succinic semi-aldehyde dehydrogenase	ssadh	E
Sulfamidase	SGSH	G
Sulfite oxidase	SUOX	E
Superoxide dismutase 1	SOD1	E
Superoxide dismutase 3	SOD3	E
Surfeit 1	SURF1	G
Survival of motor neuron 1, telomeric	SMN1	T
Synapsin 1a & 1b	SYN1	N
Synapsin 2a & 2b	SYN2	N
Synaptic vesicle amine transporter	SVAT	N
Synaptic vesicle protein 2	SV2	N
Synaptobrevin 1	SYB1	N
Synaptobrevin 2	SYB2	N
Synaptogyrin		N
Synaptophysin	SYP	N
Synaptosomal-associated protein, 25KD	SNAP25	N
Synaptotagmin 1	SYT1	N
Synaptotagmin 2	SYT2	N
Syntaxin 1	STX1	N
Tachykinin receptor, NK1R	TACR1	N
Tachykinin receptor, NK2R	TACR2	N

Tachykinin receptor, NK3R	TACR3	N
Talin	TLN	G
Tau protein	MAPT	S
TEK, tyrosine kinase, endothelial	TEK	E
Telomerase protein component		E
Thiolase, peroxisomal		E
Thrombin receptor	F2R	I
Thrombopoietin	THPO	G
Thromboxane A synthase 1	TBXAS1	I
Thromboxane A2	TXA2	I
Thromboxane A2 receptor	TBXA2R	I
Thy-1 T-cell antigen	THY1	I
Thyroxin-binding globulin	TBG	T
Tocopherol (alpha) transfer protein	TTPA	T
Topoisomerase I		E
Torticollis, keloids, cryptorchidism and renal dysplasia gene	TKCR	G
Transacylase		E
Transferrin receptor	TFRC	G
Transforming growth factor, beta 2	TGFB2	G
Transforming growth factor, beta induced	TGFB1	G
Transforming growth factor, beta receptor 2	TGFBR2	G
Transketolase-like 1	TKTL1	E
Transthyretin	TTR	T
Tremor, essential 1	ETM1	N
Tremor, essential 2	ETM2	N
Triosephosphate isomerase	TPI1	E
Tropomyosin 3 (non-muscle)	TPM3	S
Tryptophan hydroxylase	TPH	E
Tubby-like protein 1	TULP1	G
Tuberous sclerosis 1	TSC1	G
Tuberous sclerosis 2	TSC2	G
Tumour necrosis factor (TNF) receptor associated factor 1	TRAF1	I
Tumour necrosis factor (TNF) receptor associated factor 2	TRAF2	I
Tumour necrosis factor (TNF) receptor associated factor 3	TRAF3	I
Tumour necrosis factor (TNF) receptor associated factor 4	TRAF4	I
Tumour necrosis factor (TNF) receptor associated factor 5	TRAF5	I
Tumour necrosis factor (TNF) receptor associated factor 6	TRAF6	I
Tumour necrosis factor alpha	TNFA	I
Tumour necrosis factor alpha receptor	TNFAR	I
Tumour necrosis factor beta	TNFB	I
Tumour necrosis factor beta receptor	TNFBR	I



Tumour protein p53	TP53, P53	G
Tumour protein p73	TP73	G
Tyrosine aminotransferase	TAT	E
Tyrosine hydroxylase	TH	E
Ubiquitin		G
Ubiquitin B	UBB	G
Ubiquitin C	UBC	G
Ubiquitin carboxyl-terminal esterase L1	UCHL1	G
UDP-glucuronosyltransferase 1	ugt1d, UGT1	E
UDP-glucuronosyltransferase 2	UGT2	E
Urate oxidase	UOX	E
Uridinediphosphate(UDP)-galactose-4-epimerase	GALE	E
Uroporphyrinogen III synthase	UROS	E
Usher syndrome 2A	USH2A	S
Vacuolar proton pump, subunit 1	VPP1	N
Vacuolar proton pump, subunit 3	VPP3	N
Vasoactive intestinal polypeptide	VIP	N
Vasoactive intestinal polypeptide receptor	VIPR	N
Vesicular monoamine transporter 1	VMAT1	N
Vesicular monoamine transporter 2	VMAT2	N
Vitamin B12-binding (R) protein		G
Von Hippel-Lindau gene	VHL	G
Wolf-Hirschhorn syndrome candidate 1 gene	WHSC1	G
Wolfram syndrome 1 gene	WFS1	S
Xanthine dehydrogenase	XDH	E
Xeroderma pigmentosum, complementation group A	XPA	E
Zinc finger protein 2	ZIC2	S

71. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 70.
72. A set according to claim 70 or 71 in which a minority of said probes for listed genes are absent.
73. A set according to claim 70 or 71 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.
74. A set according to claim 70 or 71 in which a limited number of probes are replaced by probes for non-listed genes.

75. A set of probes for a core group of genes according to any of claims 70 to 74 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.
76. A set according to any of claims 70 to 75 consisting of probes for members of a sub-group of the core group.
77. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.
78. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.
79. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.
80. A set according to claim 77 or 78 in which said substrate is a semiconductor microchip.
81. A set according to any preceding claim for use in a biological assay for detection of said gene variants.
82. A set according to any preceding claim for use in the measurement of differential gene expression levels.
83. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
84. A medical device including a set according to any of claims 70 to 82 for use in an array for detection of differential gene expression levels.
85. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 70) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 70 and 72 to 82 and relating the probe hybridisation pattern to said variations.
86. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 71) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 71 to 82 and relating the probe interaction pattern to said variations.
87. Use of a set or device according to any of claims 70 to 82 for the prognosis and management of patients suffering from or at risk of CNS dysfunction, damage or disease or experiencing the symptoms and consequences of CNS dysfunction, damage or disease.
88. Use of a set or device according to any of claims 70 to 82 for predicting likely therapeutic response and adverse events following therapeutic intervention.
89. Use of a set or device according to any of claims 70 to 82 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
90. Use of a set or device according to any of claims 70 to 82 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.

91. Use of a set or device according to any of claims 70 to 82 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
92. Use of a set or device according to any of claims 70 to 82 for the development of new strategies of therapeutic intervention and in clinical trials.
93. Use of a set or device according to any of claims 70 to 82 for construction of and generation of algorithms for patient and healthcare management.
94. Use of a set or device according to any of claims 70 to 82 for modelling or assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations
95. Use of a set or device according to any of claims 70 to 82 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
96. Use of a set or device according to any of claims 70 to 82 for predicting optimum configuration/management of thereapeutic intervention.
97. A method according to claim 85 or 86 in which the identification of gene variants is indicative of a higher risk of developing CNS dysfunction, damage or disease or experiencing the symptoms and consequences of CNS dysfunction, damage or disease for the patient or individual.
98. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop CNS dysfunction, damage or disease or experiencing the symptoms and consequences of CNS dysfunction, damage or disease, which method comprises:
  - i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from CNS dysfunction, damage or disease;
  - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the CNS dysfunction, damage or disease;
  - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 70 to 76;
  - iv) calculating the frequencies of these alleles in the samples from i) and ii);
  - v) comparing the frequencies of these alleles in i) and ii);
  - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing symptoms and consequences of CNS dysfunction, damage or disease.
99. A method for assessing whether a given subject will be at risk of developing the symptoms and consequences of CNS dysfunction, damage or disease, which comprises comparing said subject's genotype with a model generated by the method of claim 98.
100. A method according to any of claims 85, 86, 98 and 99 wherein at least one step is computer-controlled.
101. An assay suitable for use in a method according to any of claims 85, 86, 98 and 99; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 70 to 76 in a biological sample.
102. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing the symptoms and consequences of CNS dysfunction, damage or disease; said kit comprising:

- i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 70 or 72 to 76 in a sample of human DNA
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing the symptoms and consequences of CNS dysfunction, damage or disease.
103. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing the symptoms and consequences of CNS dysfunction, damage or disease; said kit comprising:
- i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core group of genes as defined in any of claims 71 to 76 in an expressed-protein-containing human sample;
  - ii) reagents for use in the detection process readout indicating the probability of a patient or individual developing the symptoms and consequences of CNS dysfunction, damage or disease.
104. A set of probes according to claim 70, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.
105. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to behavioural disturbance and aggression; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

#### KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME  
 T TRANSPORT & STORAGE  
 S STRUCTURAL  
 I IMMUNITY  
 N NERVOUS TRANSMISSION  
 G GROWTH & DIFFERENTIATION

#### BEHAVIOUR GENE LIST

	HUGO gene symbol	Protein function
11beta hydroxysteroid dehydrogenase 2	HSD11B2	E
4-hydroxyphenylpyruvate dioxygenase	HPD	E
Acetylcholine receptor, nicotinic, alpha A1	CHRNA1	N
Acetylcholine receptor, nicotinic, alpha A2	CHRNA2	N
Acetylcholine receptor, nicotinic, alpha A3	CHRNA3	N
Acetylcholine receptor, nicotinic, alpha A4	CHRNA4	N
Acetylcholine receptor, nicotinic, alpha A5	CHRNA5	N

Acetylcholine receptor, nicotinic, alpha A6	CHRNA6	N
Acetylcholine receptor, nicotinic, alpha A7	CHRNA7	N
Acetylcholine receptor, nicotinic, beta 1	CHRNA1	N
Acetylcholine receptor, nicotinic, beta 2	CHRNA2	N
Acetylcholine receptor, nicotinic, beta 3	CHRNA3	N
Acetylcholine receptor, nicotinic, beta 4	CHRNA4	N
Acetylcholine receptor, nicotinic, epsilon	CHRNAE	N
Acetylcholine receptor, nicotinic, gamma	CHRNA6	N
Acetylcholinesterase	ACHE	E
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
alpha-synuclein	SNCA	N
Amyloid beta A4 precursor protein	APP	N
Amyloid beta A4 precursor-like protein	APLP	N
Androgen binding protein	ABP	T
Androgen receptor	AR	G
Apolipoprotein E	APOE	T
Arginosuccinate synthetase	ASS	E
Ataxia telangiectasia gene, AT	ATM	G
beta-synuclein	SNCB	N
Ca(2+) transporting ATPase, slow twitch	ATP2A2	T
Cannabinoid receptor	CNR1	N
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Catechol-O-methyltransferase	COMT	E
Cholecystokinin	CCK	N
Cholecystokinin B receptor	CCKBR	N
Choline acetyltransferase	CHAT	E
Ciliary neurotrophic factor (CNTF)	CNTF	G
Ciliary neurotrophic factor (CNTF) receptor	CNTFR	G
Corticotrophin-releasing hormone	CRH	T
Corticotrophin-releasing hormone receptor	CRHR1	T
Cryptochrome 1	CRY1	S
Cryptochrome 2	CRY2	S
Cu2+ transporting ATPase beta polypeptide	ATP7B	E
Cyclic AMP-dependent protein kinase	PKA	E
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
CYP11A1	CYP11A1	E

CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E
CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E
CYP2C18	CYP2C18	E
CYP2C19	CYP2C19	E
CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E
CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E
CYP2F1	CYP2F1	E
CYP2J2	CYP2J2	E
CYP3A3	CYP3A3	E
CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystathionase	CTH	E
Cystathione beta synthase	CBS	E
Cytidine deaminase	CDA	E
Cytidine-5-prime-triphosphate synthetase	CTPS	E
Cytochrome a		E
Cytochrome c		E
Cytochrome c oxidase, MTCO		E
Dihydrolipoamide branched chain transacylase	DBT	N
Dopamine beta hydroxylase	DBH	E
Dopamine receptors D1	DRD1	N
Dopamine receptors D2	DRD2	N

Dopamine receptors D3	DRD3	N
Dopamine receptors D4	DRD4	N
Dopamine receptors D5	DRD5	N
Doublecortin, DCX	DCX	S
Enolase	ENO1	E
Flightless-II, Drosophila homolog of	FLII	G
Fragile site, folic acid type, rare, fra(X) A	FRAXA	N
Fragile site, folic acid type, rare, fra(X) E	FRAXE	N
Fragile site, folic acid type, rare, fra(X) F	FRAXF	N
GABA receptor, alpha 1	GABRA1	N
GABA receptor, alpha 2	GABRA2	N
GABA receptor, alpha 3	GABRA3	N
GABA receptor, alpha 4	GABRA4	N
GABA receptor, alpha 5	GABRA5	N
GABA receptor, alpha 6	GABRA6	N
GABA receptor, beta 1	GABRB1	N
GABA receptor, beta 2	GABRB2	N
GABA receptor, beta 3	GABRB3	N
GABA receptor, gamma 1	GABRG1	N
GABA receptor, gamma 2	GABRG2	N
GABA receptor, gamma 3	GABRG3	N
Galactose 1-phosphate uridyl-transferase	GALT	E
Geniospasm 1	GSM1	G
Glutathione	GSH	T
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
Glycerol kinase	GK	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
GM2 ganglioside activator protein, GM2A	GM2A	E
Gustducin, alpha (taste-specific G protein)	GDCA	N
Inositol monophosphatase	IMPA1	N
IP3 kinase		E
Mannosidase, beta A lysosomal	MANBA	E
Melatonin receptor 1A	MTNR1A	N
Melatonin receptor 1B	MTNR1B	N
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
N-acetylglucosamine-6-sulfatase	GNS	E
NADPH-dependent cytochrome P450 reductase	POR	E
Neurokinin A	NKNA	N

Neurokinin B	NKNB	N
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Neurotensin	NTS	N
Neurotensin receptor	NTSR1	N
Nitric oxide synthase 1, NOS1	NOS1	E
Nitric oxide synthase 2, NOS2	NOS2	E
Nitric oxide synthase 3, NOS3	NOS3	E
Ocular albinism 1	OA1	S
Opioid receptor, delta	OPRD1	N
Opioid receptor, kappa	OPRK1	N
Opioid receptor, mu	OPRM1	N
Orexin	OX	G
Orexin 1 receptor	OX1R	G
Orexin 2 receptor	OX2R	G
Phosphoglycerate kinase 1	PGK1	E
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium voltage-gated channel E1	KCNE1	N
Potassium voltage-gated channel Q1	KCNQ1	N
Preproenkephalin	PENK	N
Preproglucagon	GCG;GLP1; GLP2	G
Prion protein	PRNP	N
Proline dehydrogenase	PRODH	E
Pro-melanin-concentrating hormone	PMCH	G
Proopiomelanocortin	POMC	N
Purine nucleoside phosphorylase	NP	E
RIGUI	RIGUI	G
Serotonin N-acetyltransferase	SNAT	E
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Solute carrier family 18, member 3	SLC18A3	T
Solute carrier family 6 (GAMMA-AMINO-BUTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T



Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Solute carrier family 6 (neurotransmitter transporter, serotonin), member 4	SLC6A4	T
Synapsin 1a & 1b	SYN1	N
Synapsin 2a & 2b	SYN2	N
Synaptogyrin		N
Synaptophysin	SYP	N
Synaptosomal-associated protein, 25KD	SNAP25	N
Syntaxin 1	STX1	N
Tachykinin receptor, NK1R	TACR1	N
Tachykinin receptor, NK2R	TACR2	N
Tachykinin receptor, NK3R	TACR3	N
Tau protein	MAPT	S
Tryptophan hydroxylase	TPH	E
Tyrosine hydroxylase	TH	E
Ubiquitin		G
Ubiquitin B	UBB	G
Ubiquitin C	UBC	G
UDP-glucuronosyltransferase 1	ugt1d, UGT1	E
UDP-glucuronosyltransferase 2	UGT2	E
Vacuolar proton pump, subunit 1	VPP1	N
Vacuolar proton pump, subunit 3	VPP3	N
Vasoactive intestinal polypeptide	VIP	N
Vasoactive intestinal polypeptide receptor	VIPR	N

106. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 105.

107. A set according to claim 105 or 106 in which a minority of said probes for listed genes are absent.

108. A set according to claim 105 or 106 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.

109. A set according to claim 105 or 106 in which a limited number of probes are replaced by probes for non-listed genes.

110. A set of probes for a core group of genes according to any of claims 105 to 109 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.

111. A set according to any of claims 105 to 110 consisting of probes for members of a sub-group of the core group.
112. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.
113. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.
114. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.
115. A set according to claim 112 or 113 in which said substrate is a semiconductor microchip.
116. A set according to any preceding claim for use in a biological assay for detection of said gene variants.
117. A set according to any preceding claim for use in the measurement of differential gene expression levels.
118. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
119. A medical device including a set according to any of claims 105 to 117 for use in an array for detection of differential gene expression levels.
120. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 105) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 105 and 107 to 117 and relating the probe hybridisation pattern to said variations.
121. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 106) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 106 to 117 and relating the probe interaction pattern to said variations.
122. Use of a set or device according to any of claims 105 to 117 for the prognosis and management of patients suffering from or at risk of developing aggressive symptoms or behavioural disturbance.
123. Use of a set or device according to any of claims 105 to 117 for predicting likely therapeutic response and adverse events following therapeutic intervention.
124. Use of a set or device according to any of claims 105 to 117 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
125. Use of a set or device according to any of claims 105 to 117 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
126. Use of a set or device according to any of claims 105 to 117 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.

127. Use of a set or device according to any of claims 105 to 117 for the development of new strategies of therapeutic intervention and in clinical trials.
128. Use of a set or device according to any of claims 105 to 117 for construction of and generation of algorithms for patient and healthcare management.
129. Use of a set or device according to any of claims 105 to 117 for modelling or assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations
130. Use of a set or device according to any of claims 105 to 117 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
131. Use of a set or device according to any of claims 105 to 117 for predicting optimum configuration/management of thereapeutic intervention.
132. A method according to claim 120 or 121 in which the identification of gene variants is indicative of a higher risk of developing aggressive symptoms and/or behavioural disturbance for the patient or individual.
133. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop aggressive symptoms and/or behavioural disturbance which method comprises:
  - i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from aggressive symptoms and/or behavioural disturbance;
  - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from aggressive symptoms and/or behavioural disturbance;
  - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 105 to 111;
  - iv) calculating the frequencies of these alleles in the samples from i) and ii);
  - v) comparing the frequencies of these alleles in i) and ii);
  - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing aggressive symptoms and/or behavioural disturbance.
134. A method for assessing whether a given subject will be at risk of developing aggressive symptoms and/or behavioural disturbance, which comprises comparing said subject's genotype with a model generated by the method of claim 133.
135. A method according to any of claims 120, 121, 133and 134 wherein at least one step is computer-controlled.
136. An assay suitable for use in a method according to any of claims 120, 121, 133 and 134; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 105 to 111 in a biological sample.
137. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing aggressive symptoms and/or behavioural disturbance; said kit comprising:
  - i) means for testing for the presence or absence or DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 105 or 107 to 111 in a sample of human DNA;

- ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing aggressive symptoms and/or behavioural disturbance.
138. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing aggressive symptoms and/or behavioural disturbance; said kit comprising:
- i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core group of genes as defined in any of claims 106 to 111 in an expressed-protein-containing human sample;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing aggressive symptoms and/or behavioural disturbance.
139. A set of probes according to claim 105, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.
140. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to brain injury; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

#### KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME  
 T TRANSPORT & STORAGE  
 S STRUCTURAL  
 I IMMUNITY  
 N NERVOUS TRANSMISSION  
 G GROWTH & DIFFERENTIATION

BRAIN INJURY GENE LIST	HUGO gene symbol	Protein function
2,3-bisphosphoglycerate mutase	BPGM	E
3 beta hydroxysteroid dehydrogenase 2	HSD3B2	E
4-hydroxyphenylpyruvate dioxygenase	HPD	E
5,10-methylenetetrahydrofolate reductase (NADPH)	MTHFR	E
6-pyruvoyltetrahydropterin synthase	PTS	E
Acetoacetyl 2-CoA-thiolase	ACAT2	E
Acetyl CoA acyltransferase	ACAA	E
Acetylcholine receptor, nicotinic, alpha A1	CHRNA1	N
Acetylcholine receptor, nicotinic, alpha A2	CHRNA2	N
Acetylcholine receptor, nicotinic, alpha A3	CHRNA3	N
Acetylcholine receptor, nicotinic, alpha A4	CHRNA4	N

Acetylcholine receptor, nicotinic, alpha A5	CHRNA5	N
Acetylcholine receptor, nicotinic, alpha A6	CHRNA6	N
Acetylcholine receptor, nicotinic, alpha A7	CHRNA7	N
Acetylcholine receptor, nicotinic, beta 1	CHRNA1	N
Acetylcholine receptor, nicotinic, beta 2	CHRNA2	N
Acetylcholine receptor, nicotinic, beta 3	CHRNA3	N
Acetylcholine receptor, nicotinic, beta 4	CHRNA4	N
Acetylcholine receptor, nicotinic, epsilon	CHRNAE	N
Acetylcholine receptor, nicotinic, gamma	CHRNA7	N
Acetylcholinesterase	ACHE	E
Adducin, alpha	ADD1	S
Adducin, beta	ADD2	S
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N
Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
Adrenocorticotrophic hormone (ACTH) receptor	ACTHR	G
Albumin, ALB	ALB	T
Aldehyde dehydrogenase 10	ALDH10	E
Aldosterone receptor	MLR	G
Alpha 1 acid glycoprotein	AAG; AGP	T
Alpha 2 macroglobulin	A2M	I
alpha thalassemia gene	ATRX	N
alpha1-antitrypsin	PI	E
alpha2-antiplasmin	PLI	E
alpha-synuclein	SNCA	N
Aminomethyltransferase	AMT	E
Aminopeptidase P	XPNPEP2	E
Amyloid beta (A4) precursor protein-binding, APBB1	APBB1	N
Amyloid beta A4 precursor protein	APP	N
Amyloid beta A4 precursor-like protein	APLP	N
Angiotensin 1	ANGPT1	G

Angiopoietin 2	ANGPT2	G
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensin receptor 1	AGTR1	T
Angiotensin receptor 2	AGTR2	T
Angiotensinogen	AGT	E
Annexin 1	ANX 1	I
Antidiuretic hormone receptor	ADHR	T
Antithrombin III	AT3	E
Apolipoprotein A I	APOA1	T
Apolipoprotein A II	APOA2	T
Apolipoprotein B	APOB	T
Apolipoprotein C1	APOC1	T
Apolipoprotein C2	APOC2	T
Apolipoprotein C3	APOC3	T
Apolipoprotein D	APOD	T
Apolipoprotein E	APOE	T
Apolipoprotein H	APOH	T
Apoptosis antigen 1	APT1	I
Arginase	ARG1	E
Arginine vasopressin	AVP	N
Arginine vasopressin receptor 1A	AVPR1A	N
Arginine vasopressin receptor 1B	AVPR1B	N
Arginine vasopressin receptor 2	AVPR2	N
Arginosuccinate lyase	ASL	E
Arginosuccinate synthetase	ASS	E
Arylsulfatase A	ARSA	E
Arylsulfatase D	ARSD	E
Arylsulfatase E	ARSE	E
Arylsulfatase F	ARSF	E
Aspartoacylase	ASPA	E
Ataxia telangiectasia gene, AT	ATM	G
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G
Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
Bagpipe homeobox, drosophila homolog of, 1	BAPX1	G
beta-synuclein	SNCB	N
Bleomycin hydrolase	BLMH	E
Bradykinin receptor B1		I
Bradykinin receptor B2		I
Brain derived neurotrophic factor	BDNF	G
Brain derived neurotrophic factor (BDNF) receptor	BDNFR	G
Butyrylcholinesterase	BCHE	E
Ca(2+) transporting ATPase, slow twitch	ATP2A2	T
Cadherin E	CDH1	G
Cadherin EP		G
Cadherin N	CDH2	G

Cadherin P	CDH3	G
Calbindin 1	CALB1	G
Calbindin D9K	CALB3	G
Calcineurin A1	CALNA1	I
Calcineurin A2	CALNA2	I
Calcineurin A3	CALNA3	I
Calcineurin B		I
Calcitonin/Calcitonin gene-related peptide alpha	CALCA	N
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha- 1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha- 1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha- 1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha- 1E (CACNL1A6)	CACNA1E	N
Calcium channel, voltage-dependent, Alpha- 2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, L type, alpha 1S subunit	CACNA1S	N
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N
Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	CACNA1A	N
Calcium channel, voltage-dependent, T-type		N
Calmodulin 1	CALM1	G
Calmodulin 2	CALM2	G
Calmodulin 3	CALM3	G
Calmodulin-dependant protein kinase II	CAMK2A	G
Calnexin	CANX	G
Calpain	CAPN, CAPN3	E
Calretinin	CALB2	N
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Cardiac-specific homeobox, CSX	CSX	G
Carnosinase		N
Caspase 1	CASP1	G
Caspase 10	CASP10	G
Caspase 2	CASP2	G
Caspase 3	CASP3	G
Caspase 4	CASP4	G

Caspase 5	CASP5	G
Caspase 6	CASP6	G
Caspase 7	CASP7	G
Caspase 8	CASP8	G
Caspase 9	CASP9	G
Catechol-O-methyltransferase	COMT	E
CD1	CD1	I
CD4	CD4	I
Cell adhesion molecule, intercellular, ICAM	ICAM1	G
Cell adhesion molecule, leukocyte- endothelial, LECAM (CD62)	LECAM1	G
Cell adhesion molecule, liver, LCAM	LCAM	G
Cell adhesion molecule, neural, NCAM1	NCAM1	G
Cell adhesion molecule, neural, NCAM120	NCAM120	G
Cell adhesion molecule, neural, NCAM2	NCAM2	G
Cell adhesion molecule, platelet-endothelial, PECAM	PECAM1	G
Cell adhesion molecule, vascular, VCAM	VCAM1	G
Ceroid lipofuscinosis neuronal 2	CLN2	N
Ceroid lipofuscinosis neuronal 3	CLN3	N
Ceroid lipofuscinosis neuronal 4	CLN4	N
Ceroid lipofuscinosis neuronal 5	CLN5	N
Ceroid lipofuscinosis neuronal 6	CLN6	N
Chemokine receptor CXCR4	CXCR4	I
Choline acetyltransferase	CHAT	E
Chymotrypsinogen		E
Cockayne syndrome gene, CKN1	CKN1	G
Cofilin		S
Collagen I alpha 1	COL1A1	S
Collagen I alpha 2	COL1A2	S
Collagen II alpha 1	COL2A1	S
Collagen III alpha 1	COL3A1	S
Collagen IV alpha 1	COL4A1	S
Collagen IV alpha 2	COL4A2	S
Collagen IV alpha 3	COL4A3	S
Collagen IV alpha 4	COL4A4	S
Collagen IV alpha 5	COL4A5	S
Collagen IV alpha 6	COL4A6	S
Collagen IX alpha 2	COL9A2, EDM2	S
Collagen IX alpha 3	COL9A3	S
Collagen receptor	COLR	S
Collagen V alpha 1	COL5A1	S
Collagen V alpha 2	COL5A2	S
Collagen VI alpha 1	COL6A1	S
Collagen VI alpha 2	COL6A2	S
Collagen VI alpha 3	COL6A3	S
Collagen VII alpha 1	COL7A1	S
Collagen X alpha 1	COL10A1	S



Collagen X alpha 1	COL11A1	S
Collagen XI alpha 2	COL11A2	S
Collagen XVII alpha 1	COL17A1	S
Corticotrophin-releasing hormone	CRH	T
Corticotrophin-releasing hormone receptor	CRHR1	T
Creb binding protein	CREBBP	G
Cu <sup>2+</sup> transporting ATPase beta polypeptide	ATP7B	E
Cyclic AMP-dependent protein kinase	PKA	E
Cyclic nucleotide phosphodiesterase 2A3	PDE2A3	E
Cyclic nucleotide phosphodiesterase 3A	PDE3A	E
Cyclic nucleotide phosphodiesterase 3B	PDE3B	E
Cyclic nucleotide phosphodiesterase 4A	PDE4A	E
Cyclic nucleotide phosphodiesterase 4C	PDE4C	E
Cyclic nucleotide phosphodiesterase 5A	PDE5A	E
Cyclic nucleotide phosphodiesterase 6A	PDE6A	E
Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E
Cyclic nucleotide phosphodiesterase 8	PDE8	E
Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
CYP11A1	CYP11A1	E
CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E
CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E
CYP2C18	CYP2C18	E
CYP2C19	CYP2C19	E
CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E
CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E
CYP2F1	CYP2F1	E
CYP2J2	CYP2J2	E
CYP3A3	CYP3A3	E

CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystatin B	CSTB	T
Cystatin C	CST3	T
Cytidine-5-prime-triphosphate synthetase	CTPS	E
Cytochrome a		E
Cytochrome c		E
Cytochrome c oxidase, MTCO		E
Cytokine-suppressive antiinflammatory drug-binding protein 1	CSBP1	I
Cytokine-suppressive antiinflammatory drug-binding protein 2	CSBP2	I
DAX1 nuclear receptor	DAX1	I
Deleted in malignant brain tumours 1	DMBT1	G
Delta-7-dehydrocholesterol reductase	DHCR7	E
Dihydrolipoamide branched chain transacylase	DBT	N
Dihydroxyacetonephosphate acyltransferase	DHAPAT	E
Dopamine beta hydroxylase	DBH	E
Dopamine receptors D1	DRD1	N
Dopamine receptors D2	DRD2	N
Dopamine receptors D3	DRD3	N
Dopamine receptors D4	DRD4	N
Dopamine receptors D5	DRD5	N
Dystonia 9	CSE	S
Dystrophin myotonia	DM, DMPK	E
Dystrophin myotonia, atypical	DM2	E
Dystrophin	DMD	S
Ectodermal Dysplasia 1 gene	ED1	S
Empty spiracles (drosophila) homologue 1	EMX1	G
Empty spiracles (drosophila) homologue 2	EMX2	G
Endothelin 1	EDN1	N
Endothelin 2	EDN2	N
Endothelin 3	EDN3	N
Endothelin converting enzyme	ECE1	N
Endothelin receptor type A	EDNRA	N
Endothelin receptor type B	EDNRB	N
Enolase	ENO1	E
Epidermal growth factor	EGF	G

Epidermal growth factor receptor	EGFR	G
Epilepsy, benign neonatal 4 gene	ICCA	E
Epilepsy, female restricted	EFMR	E
Epilepsy, progressive myoclonic 2 gene	EPM2A	E
Excision repair complementation group 4 protein	ERCC4	E
Factor 1 (No. one)	F1	I
Factor III	F3	I
Factor IX	F9	I
Factor V	F5	I
Factor VII	F7	I
Factor VIII	F8	I
Factor X	F10	I
Factor XI	F11	I
Factor XII	F12	I
Factor XIII A & B	F13A & F13B	I
Fanconi anemia, complementation group C	FANCC	T
Fanconi anemia, complementation group D	FANCD	T
Fibrinogen alpha	FGA	S
Fibrinogen beta	FGB	S
Fibrinogen gamma	FGG	S
Fibroblast growth factor	FGF1	G
Fibroblast growth factor receptor 1	FGFR1	G
Fibroblast growth factor receptor 2	FGFR2	G
Fibroblast growth factor receptor 3	FGFR3	G
Fibronectin precursor	FN1	G
Flightless-II, Drosophila homolog of	FLII	G
Follicle stimulating hormone receptor	FSHR, ODG1	G
Follicle stimulating hormone, FSH	FSHB	G
Formiminotransferase		E
Fragile site, folic acid type, rare, fra(X) A	FRAXA	N
Fragile site, folic acid type, rare, fra(X) E	FRAXE	N
Fragile site, folic acid type, rare, fra(X) F	FRAXF	N
Frataxin	FRDA	G
Fukuyama type congenital muscular dystrophy	FCMD	G
Fumarase	FH	E
GABA receptor, alpha 1	GABRA1	N
GABA receptor, alpha 2	GABRA2	N
GABA receptor, alpha 3	GABRA3	N
GABA receptor, alpha 4	GABRA4	N
GABA receptor, alpha 5	GABRA5	N
GABA receptor, alpha 6	GABRA6	N
GABA receptor, beta 1	GABRB1	N
GABA receptor, beta 2	GABRB2	N
GABA receptor, beta 3	GABRB3	N
GABA receptor, gamma 1	GABRG1	N
GABA receptor, gamma 2	GABRG2	N

GABA receptor, gamma 3	GABRG3	N
GABA transaminase	ABAT	E
Galactosyltransferase 1	GT1	G
Galactosyltransferase, alpha 1,3	GGTA1	G
Galactosyltransferase, beta 3	B3GALT	G
Galanin	GAL	N
Galanin receptor	GALNR1	N
Gamma-glutamyltransferase 1	GGT1	T
Gastric Intrinsic factor, GIF	GIF	E
GDP dissociation inhibitor 1	GDI1	G
Glial-cell derived neurotrophic factor (GDNF) receptor		N
Glial-cell derived neurotrophic factor, GDNF	GDNF	N
Glioma chloride ion channel, GCC		G
Glutamate decarboxylase, GAD	GAD1	E
Glutamate receptor 1	GLUR1	N
Glutamate receptor 2	GLUR2	N
Glutamate receptor 3	GLUR3	N
Glutamate receptor 4	GLUR4	N
Glutamate receptor 5	GLUR5	N
Glutamate receptor 6	GLUR6	N
Glutamate receptor 7	GLUR7	N
Glutamate receptor, ionotropic, NMDA 1	NMDAR1	N
Glutamate receptor, ionotropic, NMDA 2A	NMDAR2A	N
Glutamate receptor, ionotropic, NMDA 2B	NMDAR2B	N
Glutamate receptor, ionotropic, NMDA 2C	NMDAR2C	N
Glutamate receptor, ionotropic, NMDA 2D	NMDAR2D	N
Glutaryl-CoA dehydrogenase	GCDH	E
Glutathione	GSH	T
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glutathione synthetase	GSS	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
Glycerol kinase	GK	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
Glycine dehydrogenase	GLDC	E
GM2 ganglioside activator protein, GM2A	GM2A	E
Gonadotropin releasing hormone receptor	GNRHR	G
GTP cyclohydrolase 1	GCH1	G
Guanine nucleotide-binding protein, alpha activating activity polypeptide, GNAO	GNAO1	N
Guanylate cyclase 2D, membrane (retina-specific)	GUCY2D	E
Guanylate cyclase activator 1A (retina)	GUCA1A	E
Guanylyl cyclase		E
Haeme regulated inhibitor kinase		E
Haemoglobin alpha 1	HBA1	T

Haemoglobin alpha 2	HBA2	T
Haemoglobin beta	HBB	T
Haemoglobin delta	HBD	T
Haemoglobin gamma A	HBG1	T
Haemoglobin gamma B	HBG2	T
Haemoglobin gamma G	HBGG	T
Heparan sulfamidase		E
Heparin binding epidermal growth factor	HBEGF	G
Heparin Cofactor II	HCF2	I
Hepatic lipase	LIPC	E
Hexosaminidase A	HEXA,TSD	E
Hexosaminidase B	HEXB	E
Histamine receptors, H1		N
Histamine receptors, H2		N
Histamine receptors, H3		N
Histidase		E
HLA-B associated transcript 1	BAT1	I
HMG-CoA reductase	HMGCR	E
Holocarboxylase synthetase	HLCS	E
Holoprosencephaly 1	HPE1	G
Holoprosencephaly 2	HPE2	G
Holoprosencephaly 3	HPE3	G
Holoprosencephaly 4	HPE4	G
Hypoxia inducible factor 1	HIF1A	E
Hypoxia inducible factor 2		E
IC7 A and B		I
Inositol 1,4,5-triphosphate receptor 1	ITPR1	G
Inositol monophosphatase	IMPA1	N
Insulin	INS	G
Insulin receptor	INSR	G
Insulin-like growth factor 1	IGF1	G
Insulin-like growth factor 1 receptor	IGF1R	G
Insulin-like growth factor 2	IGF2	G
Insulin-like growth factor 2 receptor	IGF2R	G
Integrin beta 1	ITGB1	G
Integrin beta 2	ITGB2	G
Integrin beta 3	ITGB3	G
Integrin beta 4	ITGB4	G
Integrin beta 5	ITGB5	G
Integrin beta 6	ITGB6	G
Integrin beta 7	ITGB7	G
Integrin, alpha 1	ITGA1	G
Integrin, alpha 2	ITGA2	G
Integrin, alpha 3	ITGA3	G
Integrin, alpha 4	ITGA4	G
Integrin, alpha 5	ITGA5	G
Integrin, alpha 6	ITGA6	G
Integrin, alpha 7	ITGA7	G

Integrin, alpha 8	ITGA8	G
Integrin, alpha 9	ITGA9	G
Integrin, alpha M	ITGAM	G
Integrin, alpha X	ITGAX	G
Inter-alpha-trypsin inhibitor, IATI		E
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) 10	IL10	I
Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I
Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
IP3 kinase		E
Kallikrein 3	KAK3	I
Kininogen, High molecular weight	KNG	I
Kynureninase		E
Laminin 5, alpha 3	LAMA3	G
Laminin 5, beta 3	LAMB3	G
Laminin 5, gamma 2	LAMC2	G
Laminin M	LAMM	G
Laminin receptor 1	LAMR1	G
Latent transforming growth factor-beta binding protein 2	LTBP2	G
Leptin	LEP	G
Leptin receptor	LEPR	G
Leukin		I

Leukocyte-specific transcript 1	LST-1	I
Leukotriene A4 hydrolase		I
Leukotriene A4 synthase	LTA4S	E
Leukotriene B4 receptor		I
Leukotriene B4 synthase	LTB4S	E
Leukotriene C4 receptor		I
Leukotriene C4 synthase	LTC4S	E
Leukotriene D4/E4 receptor		I
LIM homeobox protein 1	LHX1	G
LIM-Kinase I (LINK-I)		I
Lipocortin 1	ANX4	I
Lipoprotein lipase	LPL	I
Lipoprotein receptor, Low Density	LDLR	T
Lipoprotein, High Density	HDLDT1	T
Lipoprotein, Intermediate Density		T
Lipoprotein, Low Density 1		T
Lipoprotein, Low Density 2		T
Lipoprotein, Very Low Density	VLDLR	T
Lipoprotein-associated coagulation factor	LACI	I
Low density lipoprotein receptor-related protein precursor	LRP	T
Lymphoid enhancer-binding factor	LEF-1	G
MAD (mothers against decapentaplegic, Drosophila) homologue 4	MADH4	G
Malonyl CoA decarboxylase		E
Mannosidase, alpha B lysosomal	MANB	E
Mannosidase, beta A lysosomal	MANBA	E
Methionine synthase	MTR	E
Methylmalonyl-CoA mutase	MUT	E
Mevalonate kinase	MVK	E
Mismatch repair gene, PMSL2	PMS2	G
Molybdenum cofactor synthesis 1	MOCS1	E
Molybdenum cofactor synthesis 2	MOCS2	E
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Mucopolidoses	GNPTA	E
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Myelin basic protein		S
N-acetylglucosamine-6-sulfatase	GNS	E
N-acetylglucosaminidase, alpha	NAGLU	E
NADPH-dependent cytochrome P450 reductase	POR	E
NB6		I
Nerve growth factor	NGF	G

Nerve growth factor receptor	NGFR	G
Neurite inhibitory protein		N
Neurofibromin 1	NF1	G
Neurofibromin 2	NF2	G
Neurofilament protein, NF125	NF150	S
Neurofilament protein, NF200	NF200	S
Neurofilament protein, NF68	NF68	S
Neurokinin A	NKNA	N
Neurokinin B	NKNB	N
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Nitric oxide synthase 1, NOS1	NOS1	E
Nitric oxide synthase 2, NOS2	NOS2	E
Nitric oxide synthase 3, NOS3	NOS3	E
Notch 3	NOTCH3	G
Notch ligand - jagged 1	JAG1, AGS	G
Nuclear factor I-kappa-B-like gene	IKBL	I
Nucleoside diphosphate kinase-A	NDPKA	E
Oncogene bcl2		G
Oncogene sis	PDGFB	G
Ornithine delta-aminotransferase	OAT	E
Ornithine transcarbamoylase	OTC, NME1	E
Orthodenticle (Drosophila) homolog 1	OTX1	G
Orthodenticle (Drosophila) homolog 2	OTX2	G
Patched (Drosophila) homolog, PTCH	PTCH	G
Peroxisomal membrane protein 1	PXMP1	S
Peroxisomal membrane protein 3	PXMP3	T
Peroxisome biogenesis factor 1	PEX1	T
Peroxisome biogenesis factor 19	PEX19	T
Peroxisome biogenesis factor 6	PEX6	T
Peroxisome biogenesis factor 7	PEX7	T
Peroxisome receptor 1	PXR1	T
Persyn		S
Phosphoglucose isomerase	GPI	E
Phosphoglycerate kinase 1	PGK1	E
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C alpha		I
Phospholipase C beta		I
Phospholipase C delta	PLCD1	I
Phospholipase C epsilon		I



Phospholipase C gamma	PLCG1	I
Phosphomannomutase 2	PMM2	G
Plasminogen	PLG	E
Plasminogen activator inhibitor 1	PAI1	E
Plasminogen activator inhibitor 2	PAI2	E
Plasminogen activator receptor, Urokinase	UPAR; PLAUR	S
Plasminogen activator, Tissue	PLAT; TPA	E
Plasminogen activator, Urokinase	UPA; PLAU	E
Platelet derived growth factor	PDGF	G
Platelet derived growth factor receptor	PDGFR	G
Platelet glycoprotein 1b, alpha	GP1BA	I
Platelet glycoprotein 1b, beta	GP1BB	I
Platelet glycoprotein 1b, gamma	GP1BG	I
Platelet glycoprotein IX	GP9	I
Platelet glycoprotein V	GP5	I
Platelet-activating factor acetylhydrolase 1B	PAFAH1B1 or LIS1	I
Platelet-activating factor acetylhydrolase 2	PAFAH2	I
Platelet-activating factor receptor	PAFR	I
Plectin 1	PLEC1	T
Polycystin 1	PKD1	T
Polycystin 2	PKD2	T
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium voltage-gated channel E1	KCNE1	N
Potassium voltage-gated channel Q1	KCNQ1	N
Potassium voltage-gated channel Q2	KCNQ2	N
Potassium voltage-gated channel Q3	KCNQ3	N
POU domain, class 1, transcription factor 1 (Pit1)	POU1F1	G
Prekallikrein		I
Prion protein	PRNP	N
Procollagen N-protease		E
Proline dehydrogenase	PRODH	E
Proopiomelanocortin	POMC	N
Prostacyclin synthase		I
Prostaglandin 15-OH dehydrogenase	HGPD; PGDH	I
Prostaglandin D - DP receptor		I
Prostaglandin E1 receptor		I
Prostaglandin E2 receptor		I
Prostaglandin E3 receptor		I
Prostaglandin F - FP receptor		I
Prostaglandin I2 receptor		T
Prostaglandin IP receptor		I
Protective protein for beta-galactosidase	PPGB	E
Protein C	PROC	I
Protein C inhibitor	PCI	I
Protein kinase C, alpha	PRKCA	E
Protein kinase C, gamma	PRKCG	E

Protein kinase G		E
Protein phosphatase 1, regulatory (inhibitor) subunit 3	PPP1R3	E
Protein S	PROS1	I
Prothrombin precursor	F2	I
Purine nucleoside phosphorylase	NP	E
Pyrroline-5-carboxylate synthetase	PYCS	E
Pyruvate carboxylase	PC	E
Ras-G-protein	RAS	G
Renin	REN	E
Replication factor C	RFC2	E
RIGUI	RIGUI	G
S100 calcium-binding protein A1	S100A1	N
S100 calcium-binding protein A2	S100A2	N
S100 calcium-binding protein A3	S100A3	N
S100 calcium-binding protein A4	S100A4	N
S100 calcium-binding protein A5	S100A5	N
S100 calcium-binding protein A6	S100A6	N
S100 calcium-binding protein A7	S100A7	N
S100 calcium-binding protein A8	S100A8	N
S100 calcium-binding protein A9	S100A9	N
S100 calcium-binding protein B	S100B	N
S100 calcium-binding protein P	S100P	N
Secretase, alpha		N
Secretase, beta		N
Secretase, gamma		N
Selectin E	SELE	N
Selectin L	SELL	N
Selectin P	SELP	N
Serotonin N-acetyltransferase	SNAT	E
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N

Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Solute carrier family 1 (glutamate transporter), member 1	SLC1A1	T
Solute carrier family 1 (glutamate transporter), member 2	SLC1A2	T
Solute carrier family 12, member 1	SLC12A1	T
Solute carrier family 12, member 2	SLC12A2	T
Solute carrier family 12, member 3	SLC12A3	T
Solute carrier family 16 (monocarboxylate transporter), member 1	SLC16A1	T
Solute carrier family 16 (monocarboxylate transporter), member 7	SLC16A7	T
Solute carrier family 18, member 3	SLC18A3	T
Solute carrier family 2 (facilitated glucose transporter), member 1	SLC2A1	T
Solute carrier family 20, member 3	SLC20A3	T
Solute carrier family 5 (sodium/glucose transporter), member 1	SLC5A1	T
Solute carrier family 5 (sodium/glucose transporter), member 2	SLC5A2	T
Solute carrier family 5 (sodium/glucose transporter), member 5	SLC5A5	T
Solute carrier family 5, member 3	SLC5A3	T
Solute carrier family 6 (GAMMA-AMINO BUTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Solute carrier family 6 (neurotransmitter transporter, serotonin), member 4	SLC6A4	T
Solute carrier family 7 (amino acid transporter), member 1	SLC7A1	T
Solute carrier family 7 (amino acid transporter), member 2	SLC7A2	T
Solute carrier family 7 (amino acid transporter), member 7	SLC7A7	T
Sphingomyelinase	SMPD1	E
Spinocerebellar ataxia 8 gene	SCA8	N
Steroid 5 alpha reductase 1	SRD5A1	E
Steroid 5 alpha reductase 2	SRD5A2	E
Substance P		N
Succinic semi-aldehyde dehydrogenase	ssadh	E
Sulfamidase	SGSH	G
Sulfite oxidase	SUOX	E
Superoxide dismutase 1	SOD1	E

Superoxide dismutase 3	SOD3	E
Surfeit 1	SURF1	G
Synapsin 1a & 1b	SYN1	N
Synapsin 2a & 2b	SYN2	N
Synaptic vesicle amine transporter	SVAT	N
Synaptobrevin 1	SYB1	N
Synaptobrevin 2	SYB2	N
Synaptogyrin		N
Synaptophysin	SYP	N
Synaptotagmin 1	SYT1	N
Synaptotagmin 2	SYT2	N
Syntaxin 1	STX1	N
Talin	TLN	G
Tau protein	MAPT	S
TEK, tyrosine kinase, endothelial	TEK	E
Telomerase protein component		E
Thrombin receptor	F2R	I
Thrombopoietin	THPO	G
Thromboxane A synthase 1	TBXAS1	I
Thromboxane A2	TXA2	I
Thromboxane A2 receptor	TBXA2R	I
Thyroxine-binding globulin	TBG	T
Topoisomerase I		E
Transforming growth factor, beta 2	TGFB2	G
Transforming growth factor, beta receptor 2	TGFBR2	G
Tuberous sclerosis 1	TSC1	G
Tuberous sclerosis 2	TSC2	G
Tumour necrosis factor (TNF) receptor associated factor 1	TRAF1	I
Tumour necrosis factor (TNF) receptor associated factor 2	TRAF2	I
Tumour necrosis factor (TNF) receptor associated factor 3	TRAF3	I
Tumour necrosis factor (TNF) receptor associated factor 4	TRAF4	I
Tumour necrosis factor (TNF) receptor associated factor 5	TRAF5	I
Tumour necrosis factor (TNF) receptor associated factor 6	TRAF6	I
Tumour necrosis factor alpha	TNFA	I
Tumour necrosis factor alpha receptor	TNFAR	I
Tumour necrosis factor beta	TNFB	I
Tumour necrosis factor beta receptor	TNFBR	I
Tumour protein p53	TP53, P53	G
Tumour protein p63	TP63	G
Tyrosine aminotransferase	TAT	E
Tyrosine hydroxylase	TH	E
Ubiquitin		G

Ubiquitin B	UBB	G
Ubiquitin C	UBC	G
Ubiquitin carboxyl-terminal esterase L1	UCHL1	G
UDP-glucuronosyltransferase 1	ugt1d, UGT1	E
UDP-glucuronosyltransferase 2	UGT2	E
Undulin 1	COL14A1	S
Uridinediphosphate(UDP)-galactose-4-epimerase	GALE	E
Uroporphyrinogen III synthase	UROS	E
Vacuolar proton pump, subunit 1	VPP1	N
Vacuolar proton pump, subunit 3	VPP3	N
Vasoactive intestinal polypeptide	VIP	N
Vasoactive intestinal polypeptide receptor	VIPR	N
Von Hippel-Lindau gene	VHL	G
Wolf-Hirschhorn syndrome candidate 1 gene	WHSC1	G
Xanthine dehydrogenase	XDH	E
Zinc finger protein 2	ZIC2	S

141. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 140.
142. A set according to claim 140 or 141 in which a minority of said probes for listed genes are absent.
143. A set according to claim 140 or 141 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.
144. A set according to claim 140 or 141 in which a limited number of probes are replaced by probes for non-listed genes.
145. A set of probes for a core group of genes according to any of claims 140 to 144 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.
146. A set according to any of claims 140 to 145 consisting of probes for members of a sub-group of the core group.
147. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.
148. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.

149. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.
150. A set according to claim 147 or 148 in which said substrate is a semiconductor microchip.
151. A set according to any preceding claim for use in a biological assay for detection of said gene variants.
152. A set according to any preceding claim for use in the measurement of differential gene expression levels.
153. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
154. A medical device including a set according to any of claims 140 to 152 for use in an array for detection of differential gene expression levels.
155. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 140) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 140 and 142 to 152 and relating the probe hybridisation pattern to said variations.
156. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 141) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 141 to 152 and relating the probe interaction pattern to said variations.
157. Use of a set or device according to any of claims 140 to 152 for the prognosis and management of patients suffering from or at risk of clinical, psychological and social consequences of brain injury.
158. Use of a set or device according to any of claims 140 to 152 for predicting likely therapeutic response and adverse events following therapeutic intervention.
159. Use of a set or device according to any of claims 140 to 152 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
160. Use of a set or device according to any of claims 140 to 152 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
161. Use of a set or device according to any of claims 140 to 152 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
162. Use of a set or device according to any of claims 140 to 152 for the development of new strategies of therapeutic intervention and in clinical trials.
163. Use of a set or device according to any of claims 140 to 152 for construction of and generation of algorithms for patient and healthcare management.
164. Use of a set or device according to any of claims 140 to 152 for modelling or assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations

165. Use of a set or device according to any of claims 140 to 152 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
166. Use of a set or device according to any of claims 140 to 152 for predicting optimum configuration/management of thereapeutic intervention.
167. A method according to claim 155 or 156 in which the identification of gene variants is indicative of a higher risk of developing clinical, psychological and social consequences of brain injury for the patient or individual.
168. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop clinical, psychological and social consequences of brain injury, which method comprises:
  - i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from clinical, psychological and social consequences of brain injury;
  - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the clinical, psychological and social consequences of brain injury;
  - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 140 to 146;
  - iv) calculating the frequencies of these alleles in the samples from i) and ii);
  - v) comparing the frequencies of these alleles in i) and ii);
  - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing clinical, psychological and social consequences of brain injury.
169. A method for assessing whether a given subject will be at risk of developing clinical, psychological and social consequences of brain injury, which comprises comparing said subject's genotype with a model generated by the method of claim 168.
170. A method according to any of claims 155, 156, 168 and 169 wherein at least one step is computer-controlled.
171. An assay suitable for use in a method according to any of claims 155, 156, 168 and 169; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 140 to 146 in a biological sample.
172. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing clinical, psychological and social consequences of brain injury; said kit comprising:
  - i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 140 or 142 to 146 in a sample of human DNA;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing clinical, psychological and social consequences of brain injury.
173. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing clinical, psychological and social consequences of brain injury; said kit comprising:

- i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core group of genes as defined in any of claims 141 to 146 in an expressed-protein-containing human sample;
- ii) reagents for use in the detection process
- iii) readout indicating the probability of a patient or individual developing clinical, psychological and social consequences of brain injury.

174. A set of probes according to claim 140, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.

175. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to dementia and/or its associated symptoms; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

#### KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME  
 T TRANSPORT & STORAGE  
 S STRUCTURAL  
 I IMMUNITY  
 N NERVOUS TRANSMISSION  
 G GROWTH & DIFFERENTIATION

#### DEMENTIA GENE LIST

	HUGO gene symbol	Protein function
2,3-bisphosphoglycerate mutase	BPGM	E
3 beta hydroxysteroid dehydrogenase 2	HSD3B2	E
5,10-methylenetetrahydrofolate reductase (NADPH)	MTHFR	E
Acetylcholine receptor, nicotinic, alpha A1	CHRNA1	N
Acetylcholine receptor, nicotinic, alpha A2	CHRNA2	N
Acetylcholine receptor, nicotinic, alpha A3	CHRNA3	N
Acetylcholine receptor, nicotinic, alpha A4	CHRNA4	N
Acetylcholine receptor, nicotinic, alpha A5	CHRNA5	N
Acetylcholine receptor, nicotinic, alpha A6	CHRNA6	N
Acetylcholine receptor, nicotinic, alpha A7	CHRNA7	N
Acetylcholine receptor, nicotinic, beta 1	CHRNA1	N
Acetylcholine receptor, nicotinic, beta 2	CHRNA2	N
Acetylcholine receptor, nicotinic, beta 3	CHRNA3	N
Acetylcholine receptor, nicotinic, beta 4	CHRNA4	N
Acetylcholine receptor, nicotinic, epsilon	CHRNA5	N
Acetylcholine receptor, nicotinic, gamma	CHRNA6	N
Acetylcholinesterase	ACHE	E



Adducin, alpha	ADD1	S
Adducin, beta	ADD2	S
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N
Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
Adrenocorticotrophic hormone (ACTH) receptor	ACTHR	G
Albumin, ALB	ALB	T
Aldosterone receptor	MLR	G
Alpha 2 macroglobulin	A2M	I
alpha1-antitrypsin	PI	E
alpha2-antiplasmin	PLI	E
alpha-synuclein	SNCA	N
Aminopeptidase P	XPNPEP2	E
Amyloid beta (A4) precursor protein-binding, APBB1	APBB1	N
Amyloid beta A4 precursor protein	APP	N
Amyloid beta A4 precursor-like protein	APLP	N
Angiopoietin 1	ANGPT1	G
Angiopoietin 2	ANGPT2	G
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensin receptor 1	AGTR1	T
Angiotensin receptor 2	AGTR2	T
Angiotensinogen	AGT	E
Antidiuretic hormone receptor	ADHR	T
Antithrombin III	AT3	E
Apolipoprotein A I	APOA1	T
Apolipoprotein A II	APOA2	T
Apolipoprotein B	APOB	T
Apolipoprotein C1	APOC1	T
Apolipoprotein C2	APOC2	T
Apolipoprotein C3	APOC3	T
Apolipoprotein D	APOD	T

Apolipoprotein E	APOE	T
Apolipoprotein H	APOH	T
Apoptosis antigen 1	APT1	I
Arginase	ARG1	E
Arginine vasopressin	AVP	N
Arginine vasopressin receptor 1A	AVPR1A	N
Arginine vasopressin receptor 1B	AVPR1B	N
Arginine vasopressin receptor 2	AVPR2	N
Arginosuccinate lyase	ASL	E
Arginosuccinate synthetase	ASS	E
Ataxia telangiectasia gene, AT	ATM	G
ATP/ADP translocase		E
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G
Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
Bagpipe homeobox, drosophila homolog of, 1	BAPX1	G
beta-synuclein	SNCB	N
Bleomycin hydrolase	BLMH	E
Bradykinin receptor B1		I
Bradykinin receptor B2		I
Brain derived neurotrophic factor	BDNF	G
Brain derived neurotrophic factor (BDNF) receptor	BDNFR	G
Butyrylcholinesterase	BCHE	E
Cadherin E	CDH1	G
Cadherin EP		G
Cadherin N	CDH2	G
Cadherin P	CDH3	G
Calbindin 1	CALB1	G
Calbindin D9K	CALB3	G
Calcineurin A1	CALNA1	I
Calcineurin A2	CALNA2	I
Calcineurin A3	CALNA3	I
Calcineurin B		I
Calcitonin/Calcitonin gene-related peptide alpha	CALCA	N
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha-1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha-1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha-1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha-1E (CACNL1A6)	CACNA1E	N
Calcium channel, voltage-dependent, Alpha-	CACNA2	N

## 2/delta

Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, L type, alpha 1S subunit	CACNA1S	N
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N
Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	CACNA1A	N
Calcium channel, voltage-dependent, T-type		N
Calmodulin 1	CALM1	G
Calmodulin 2	CALM2	G
Calmodulin 3	CALM3	G
Calmodulin-dependant protein kinase II	CAMK2A	G
Calnexin	CANX	G
Calpain	CAPN, CAPN3	E
Calretinin	CALB2	N
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Cardiac-specific homeobox, CSX	CSX	G
Caspase 1	CASP1	G
Caspase 10	CASP10	G
Caspase 2	CASP2	G
Caspase 3	CASP3	G
Caspase 4	CASP4	G
Caspase 5	CASP5	G
Caspase 6	CASP6	G
Caspase 7	CASP7	G
Caspase 8	CASP8	G
Caspase 9	CASP9	G
Catechol-O-methyltransferase	COMT	E
CD1	CD1	I
CD4	CD4	I
Cell adhesion molecule, intercellular, ICAM	ICAM1	G
Cell adhesion molecule, leukocyte-endothelial, LECAM (CD62)	LECAM1	G
Cell adhesion molecule, liver, LCAM	LCAM	G
Cell adhesion molecule, neural, NCAM1	NCAM1	G
Cell adhesion molecule, neural, NCAM120	NCAM120	G
Cell adhesion molecule, neural, NCAM2	NCAM2	G
Cell adhesion molecule, platelet-endothelial, PECAM	PECAM1	G
Cell adhesion molecule, vascular, VCAM	VCAM1	G
Chemokine receptor CXCR4	CXCR4	I
Choline acetyltransferase	CHAT	E
Chymotrypsinogen		E

Cockayne syndrome gene, CKN1	CKN1	G
Cofilin		S
Collagen I alpha 1	COL1A1	S
Collagen I alpha 2	COL1A2	S
Collagen II alpha 1	COL2A1	S
Collagen III alpha 1	COL3A1	S
Collagen IV alpha 1	COL4A1	S
Collagen IV alpha 2	COL4A2	S
Collagen IV alpha 3	COL4A3	S
Collagen IV alpha 4	COL4A4	S
Collagen IV alpha 5	COL4A5	S
Collagen IV alpha 6	COL4A6	S
Collagen IX alpha 2	COL9A2, EDM2	S
Collagen IX alpha 3	COL9A3	S
Collagen receptor	COLR	S
Collagen V alpha 1	COL5A1	S
Collagen V alpha 2	COL5A2	S
Collagen VI alpha 1	COL6A1	S
Collagen VI alpha 2	COL6A2	S
Collagen VI alpha 3	COL6A3	S
Collagen VII alpha 1	COL7A1	S
Collagen X alpha 1	COL10A1	S
Collagen X alpha 1	COL11A1	S
Collagen XI alpha 2	COL11A2	S
Collagen XVII alpha 1	COL17A1	S
Corticotrophin-releasing hormone	CRH	T
Corticotrophin-releasing hormone receptor	CRHR1	T
Cu <sup>2+</sup> transporting ATPase beta polypeptide	ATP7B	E
Cyclic AMP-dependent protein kinase	PKA	E
Cyclic nucleotide phosphodiesterase 2A3	PDE2A3	E
Cyclic nucleotide phosphodiesterase 3A	PDE3A	E
Cyclic nucleotide phosphodiesterase 3B	PDE3B	E
Cyclic nucleotide phosphodiesterase 4A	PDE4A	E
Cyclic nucleotide phosphodiesterase 4C	PDE4C	E
Cyclic nucleotide phosphodiesterase 5A	PDE5A	E
Cyclic nucleotide phosphodiesterase 6A	PDE6A	E
Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E
Cyclic nucleotide phosphodiesterase 8	PDE8	E
Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
CYP11A1	CYP11A1	E
CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E

CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E
CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E
CYP2C18	CYP2C18	E
CYP2C19	CYP2C19	E
CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E
CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E
CYP2F1	CYP2F1	E
CYP2J2	CYP2J2	E
CYP3A3	CYP3A3	E
CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystathione beta synthase	CBS	E
Cystatin C	CST3	T
Cystinosin	CTNS	T
Cytidine-5-prime-triphosphate synthetase	CTPS	E
Cytochrome a		E
Cytochrome b-245 alpha	CYBA	E
Cytochrome b-245 beta	CYBB	E
Cytochrome c		E
Cytochrome c oxidase, MTCO		E
Dihydrolipoyl succinyltransferase	DLST	E
Dopamine beta hydroxylase	DBH	E
Dopamine receptors D1	DRD1	N
Dopamine receptors D2	DRD2	N
Dopamine receptors D3	DRD3	N
Dopamine receptors D4	DRD4	N
Dopamine receptors D5	DRD5	N

Doublecortin, DCX	DCX	S
Emerin	EMD	T
Endothelin 1	EDN1	N
Endothelin 2	EDN2	N
Endothelin 3	EDN3	N
Endothelin converting enzyme	ECE1	N
Endothelin receptor type A	EDNRA	N
Endothelin receptor type B	EDNRB	N
Enolase	ENO1	E
Epidermal growth factor	EGF	G
Epidermal growth factor receptor	EGFR	G
Epilepsy, progressive myoclonic 2 gene	EPM2A	E
Excision repair complementation group 4 protein	ERCC4	E
Factor 1 (No. one)	F1	I
Factor III	F3	I
Factor IX	F9	I
Factor V	F5	I
Factor VII	F7	I
Factor VIII	F8	I
Factor X	F10	I
Factor XI	F11	I
Factor XII	F12	I
Factor XIII A & B	F13A & F13B	I
Fanconi anemia, complementation group A	FANCA	T
Fibrinogen alpha	FGA	S
Fibrinogen beta	FGB	S
Fibrinogen gamma	FGG	S
Fibroblast growth factor	FGF1	G
Fibroblast growth factor receptor 1	FGFR1	G
Fibroblast growth factor receptor 2	FGFR2	G
Fibroblast growth factor receptor 3	FGFR3	G
Fibronectin precursor	FN1	G
Follicle stimulating hormone receptor	FSHR, ODG1	G
Follicle stimulating hormone, FSH	FSHB	G
GABA receptor, alpha 1	GABRA1	N
GABA receptor, alpha 2	GABRA2	N
GABA receptor, alpha 3	GABRA3	N
GABA receptor, alpha 4	GABRA4	N
GABA receptor, alpha 5	GABRA5	N
GABA receptor, alpha 6	GABRA6	N
GABA receptor, beta 1	GABRB1	N
GABA receptor, beta 2	GABRB2	N
GABA receptor, beta 3	GABRB3	N
GABA receptor, gamma 1	GABRG1	N
GABA receptor, gamma 2	GABRG2	N
GABA receptor, gamma 3	GABRG3	N
GABA transaminase	ABAT	E

Galactosyltransferase 1	GT1	G
Galactosyltransferase, alpha 1,3	GGTA1	G
Galactosyltransferase, beta 3	B3GALT	G
Gastric Intrinsic factor, GIF	GIF	E
Glial-cell derived neurotrophic factor (GDNF) receptor		N
Glial-cell derived neurotrophic factor, GDNF	GDNF	N
Glutamate decarboxylase, GAD	GAD1	E
Glutamate receptor 1	GLUR1	N
Glutamate receptor 2	GLUR2	N
Glutamate receptor 3	GLUR3	N
Glutamate receptor 4	GLUR4	N
Glutamate receptor 5	GLUR5	N
Glutamate receptor 6	GLUR6	N
Glutamate receptor 7	GLUR7	N
Glutamate receptor, ionotropic, NMDA 1	NMDAR1	N
Glutamate receptor, ionotropic, NMDA 2A	NMDAR2A	N
Glutamate receptor, ionotropic, NMDA 2B	NMDAR2B	N
Glutamate receptor, ionotropic, NMDA 2C	NMDAR2C	N
Glutamate receptor, ionotropic, NMDA 2D	NMDAR2D	N
Glutaryl-CoA dehydrogenase	GCDH	E
Glutathione	GSH	T
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
Glycerol kinase	GK	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
Gonadotropin releasing hormone receptor	GNRHR	G
Guanylyl cyclase		E
Haemoglobin alpha 1	HBA1	T
Haemoglobin alpha 2	HBA2	T
Haemoglobin beta	HBB	T
Haemoglobin delta	HBD	T
Haemoglobin gamma A	HBG1	T
Haemoglobin gamma B	HBG2	T
Haemoglobin gamma G	HBGG	T
Heparan sulfamidase		E
Heparin binding epidermal growth factor	HBEGF	G
Heparin Cofactor II	HCF2	I
Hepatic lipase	LIPC	E
Hexosaminidase A	HEXA,TSD	E
Hexosaminidase B	HEXB	E
Hippocampal cholinergic neurostimulating peptide, HCNP		N
Histamine receptors, H1		N
Histamine receptors, H2		N
Histamine receptors, H3		N
Histidase		E

HLA-B associated transcript 1	BAT1	I
HMG-CoA reductase	HMGCR	E
Holocarboxylase synthetase	HLCS	E
Hypoxia inducible factor 1	HIF1A	E
Hypoxia inducible factor 2		E
IC7 A and B		I
Inositol monophosphatase	IMPA1	N
Insulin	INS	G
Insulin receptor	INSR	G
Integrin beta 1	ITGB1	G
Integrin beta 2	ITGB2	G
Integrin beta 3	ITGB3	G
Integrin beta 4	ITGB4	G
Integrin beta 5	ITGB5	G
Integrin beta 6	ITGB6	G
Integrin beta 7	ITGB7	G
Integrin, alpha 1	ITGA1	G
Integrin, alpha 2	ITGA2	G
Integrin, alpha 3	ITGA3	G
Integrin, alpha 4	ITGA4	G
Integrin, alpha 5	ITGA5	G
Integrin, alpha 6	ITGA6	G
Integrin, alpha 7	ITGA7	G
Integrin, alpha 8	ITGA8	G
Integrin, alpha 9	ITGA9	G
Integrin, alpha M	ITGAM	G
Integrin, alpha X	ITGAX	G
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) 10	IL10	I
Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I
Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I



Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
IP3 kinase		E
Kallikrein 3	KAK3	I
Kininogen, High molecular weight	KNG	I
Kynureninase		E
Laminin 5, alpha 3	LAMA3	G
Laminin 5, beta 3	LAMB3	G
Laminin 5, gamma 2	LAMC2	G
Laminin M	LAMM	G
Laminin receptor 1	LAMR1	G
Latent transforming growth factor-beta binding protein 2	LTBP2	G
Leptin	LEP	G
Leptin receptor	LEPR	G
Leukin		I
Leukocyte-specific transcript 1	LST-1	I
Leukotriene A4 hydrolase		I
Leukotriene A4 synthase	LTA4S	E
Leukotriene B4 receptor		I
Leukotriene B4 synthase	LTB4S	E
Leukotriene C4 receptor		I
Leukotriene C4 synthase	LTC4S	E
Leukotriene D4/E4 receptor		I
LIM homeobox protein 1	LHX1	G
LIM-Kinase I (LINK-I)		I
Lipoprotein receptor, Low Density	LDLR	T
Lipoprotein, High Density	HDLDT1	T
Lipoprotein, Intermediate Density		T
Lipoprotein, Low Density 1		T
Lipoprotein, Low Density 2		T
Lipoprotein, Very Low Density	VLDLR	T
Low density lipoprotein receptor-related protein precursor	LRP	T
Lymphoid enhancer-binding factor	LEF-1	G
MAD (mothers against decapentaplegic, Drosophila) homologue 4	MADH4	G
Mannosidase, alpha B lysosomal	MANB	E
Mannosidase, beta A lysosomal	MANBA	E
Methionine synthase	MTR	E
Mismatch repair gene, PMSL2	PMS2	G
Molybdenum cofactor synthesis 1	MOCS1	E

Molybdenum cofactor synthesis 2	MOCS2	E
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Myelin basic protein		S
N-acetylglucosamine-6-sulfatase	GNS	E
N-acetylglucosaminidase, alpha	NAGLU	E
NADPH-dependent cytochrome P450 reductase	POR	E
NB6		I
Nerve growth factor	NGF	G
Nerve growth factor receptor	NGFR	G
Neurite inhibitory protein		N
Neuroendocrine convertase 1	NEC1, PCSK1	E
Neurofibromin 1	NF1	G
Neurofibromin 2	NF2	G
Neurofilament protein, NF125	NF150	S
Neurofilament protein, NF200	NF200	S
Neurofilament protein, NF68	NF68	S
Neurokinin A	NKNA	N
Neurokinin B	NKNB	N
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Nitric oxide synthase 1, NOS1	NOS1	E
Nitric oxide synthase 2, NOS2	NOS2	E
Nitric oxide synthase 3, NOS3	NOS3	E
Notch 3	NOTCH3	G
Nuclear factor I-kappa-B-like gene	IKBL	I
Nucleoside diphosphate kinase-A	NDPKA	E
Oncogene bcl2		G
Oncogene sis	PDGFB	G
Ornithine delta-aminotransferase	OAT	E
Ornithine transcarbamoylase	OTC, NME1	E
Parkin	PARK2	N
Persyn		S
Phosphoglucose isomerase	GPI	E
Phosphoglycerate kinase 1	PGK1	E
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I

Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C alpha		I
Phospholipase C beta		I
Phospholipase C delta	PLCD1	I
Phospholipase C epsilon		I
Phospholipase C gamma	PLCG1	I
Plasminogen	PLG	E
Plasminogen activator inhibitor 1	PAI1	E
Plasminogen activator inhibitor 2	PAI2	E
Plasminogen activator receptor, Urokinase	UPAR; PLAUR	S
Plasminogen activator, Tissue	PLAT; TPA	E
Plasminogen activator, Urokinase	UPA; PLAU	E
Platelet derived growth factor	PDGF	G
Platelet derived growth factor receptor	PDGFR	G
Platelet-activating factor receptor	PAFR	I
Postsynaptic density-95 protein	PSD95	N
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium voltage-gated channel E1	KCNE1	N
Potassium voltage-gated channel Q1	KCNQ1	N
POU domain, class 1, transcription factor 1 (Pit1)	POU1F1	G
Prekallikrein		I
Presenilin 1	PSEN1	T
Presenilin 2	PSEN2	T
Prion protein	PRNP	N
Procollagen N-protease		E
Proopiomelanocortin	POMC	N
Prostacyclin synthase		I
Prostaglandin 15-OH dehydrogenase	HGPD; PGDH	I
Prostaglandin D - DP receptor		I
Prostaglandin E1 receptor		I
Prostaglandin E2 receptor		I
Prostaglandin E3 receptor		I
Prostaglandin F - FP receptor		I
Prostaglandin I2 receptor		T
Prostaglandin IP receptor		I
Protective protein for beta-galactosidase	PPGB	E
Protein C	PROC	I
Protein C inhibitor	PCI	I
Protein kinase C, alpha	PRKCA	E
Protein kinase C, gamma	PRKCG	E
Protein kinase G		E
Protein phosphatase 1, regulatory (inhibitor) subunit 3	PPP1R3	E
Protein S	PROS1	I
Prothrombin precursor	F2	I
Purine nucleoside phosphorylase	NP	E

Pyruvate carboxylase	PC	E
Renin	REN	E
Replication factor C	RFC2	E
RIGUI	RIGUI	G
S100 calcium-binding protein A1	S100A1	N
S100 calcium-binding protein A2	S100A2	N
S100 calcium-binding protein A3	S100A3	N
S100 calcium-binding protein A4	S100A4	N
S100 calcium-binding protein A5	S100A5	N
S100 calcium-binding protein A6	S100A6	N
S100 calcium-binding protein A7	S100A7	N
S100 calcium-binding protein A8	S100A8	N
S100 calcium-binding protein A9	S100A9	N
S100 calcium-binding protein B	S100B	N
S100 calcium-binding protein P	S100P	N
Secretase, alpha		N
Secretase, beta		N
Secretase, gamma		N
Selectin E	SELE	N
Selectin L	SELL	N
Selectin P	SELP	N
Serotonin N-acetyltransferase	SNAT	E
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage-gated, type 1, beta	SCN1B	N
polypeptide		
Solute carrier family 1 (glutamate transporter), member 1	SLC1A1	T
Solute carrier family 1 (glutamate transporter), member 2	SLC1A2	T
Solute carrier family 12, member 1	SLC12A1	T
Solute carrier family 12, member 2	SLC12A2	T
Solute carrier family 12, member 3	SLC12A3	T

Solute carrier family 18, member 3	SLC18A3	T
Solute carrier family 5 (sodium/glucose transporter), member 1	SLC5A1	T
Solute carrier family 5 (sodium/glucose transporter), member 2	SLC5A2	T
Solute carrier family 5 (sodium/glucose transporter), member 5	SLC5A5	T
Solute carrier family 5, member 3	SLC5A3	T
Solute carrier family 6 (GAMMA-AMINO BUTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Solute carrier family 6 (neurotransmitter transporter, serotonin), member 4	SLC6A4	T
Sphingomyelinase	SMPD1	E
Substance P		N
Succinic semi-aldehyde dehydrogenase	ssadh	E
Sulfite oxidase	SUOX	E
Superoxide dismutase 1	SOD1	E
Superoxide dismutase 3	SOD3	E
Surfeit 1	SURF1	G
Synaptogyrin		N
Synaptophysin	SYP	N
Syntaxin 1	STX1	N
Talin	TLN	G
Tau protein	MAPT	S
TEK, tyrosine kinase, endothelial	TEK	E
Telomerase protein component		E
Thrombin receptor	F2R	I
Thrombopoietin	THPO	G
Thromboxane A synthase 1	TBXAS1	I
Topoisomerase I		E
Transforming growth factor, beta 2	TGFB2	G
Transforming growth factor, beta receptor 2	TGFBR2	G
Tumour necrosis factor (TNF) receptor associated factor 1	TRAF1	I
Tumour necrosis factor (TNF) receptor associated factor 2	TRAF2	I
Tumour necrosis factor (TNF) receptor associated factor 3	TRAF3	I
Tumour necrosis factor (TNF) receptor associated factor 4	TRAF4	I
Tumour necrosis factor (TNF) receptor associated factor 5	TRAF5	I
Tumour necrosis factor (TNF) receptor associated factor 6	TRAF6	I

Tumour necrosis factor alpha	TNFA	I
Tumour necrosis factor alpha receptor	TNFAR	I
Tumour necrosis factor beta	TNFB	I
Tumour necrosis factor beta receptor	TNFBR	I
Tumour protein p53	TP53, P53	G
Tumour protein p63	TP63	G
Tyrosine aminotransferase	TAT	E
Tyrosine hydroxylase	TH	E
Ubiquitin		G
Ubiquitin B	UBB	G
Ubiquitin C	UBC	G
Ubiquitin carboxyl-terminal esterase L1	UCHL1	G
UDP-glucuronosyltransferase 1	ugt1d, UGT1	E
UDP-glucuronosyltransferase 2	UGT2	E
Uridinediphosphate(UDP)-galactose-4-epimerase	GALE	E
Uroporphyrinogen III synthase	UROS	E
Vacuolar proton pump, subunit 1	VPP1	N
Vacuolar proton pump, subunit 3	VPP3	N
Vasoactive intestinal polypeptide	VIP	N
Vasoactive intestinal polypeptide receptor	VIPR	N
Xanthine dehydrogenase	XDH	E

176. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 175.

177. A set according to claim 175 or 176 in which a minority of said probes for listed genes are absent.

178. A set according to claim 175 or 176 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.

179. A set according to claim 175 or 176 in which a limited number of probes are replaced by probes for non-listed genes.

180. A set of probes for a core group of genes according to any of claims 175 to 179 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.

181. A set according to any of claims 175 to 180 consisting of probes for members of a sub-group of the core group.

182. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.
183. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.
184. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.
185. A set according to claim 182 or 183 in which said substrate is a semiconductor microchip.
186. A set according to any preceding claim for use in a biological assay for detection of said gene variants.
187. A set according to any preceding claim for use in the measurement of differential gene expression levels.
188. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
189. A medical device including a set according to any of claims 175 to 187 for use in an array for detection of differential gene expression levels.
190. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 175) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 175 and 177 to 187 and relating the probe hybridisation pattern to said variations.
191. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 176) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 176 to 187 and relating the probe interaction pattern to said variations.
192. Use of a set or device according to any of claims 175 to 187 for the prognosis and management of patients suffering from or at risk of dementia and/or its associated symptoms.
193. Use of a set or device according to any of claims 175 to 187 for predicting likely therapeutic response and adverse events following therapeutic intervention.
194. Use of a set or device according to any of claims 175 to 187 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
195. Use of a set or device according to any of claims 175 to 187 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
196. Use of a set or device according to any of claims 175 to 187 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
197. Use of a set or device according to any of claims 175 to 187 for the development of new strategies of therapeutic intervention and in clinical trials.
198. Use of a set or device according to any of claims 175 to 187 for construction of and generation of algorithms for patient and healthcare management.
199. Use of a set or device according to any of claims 175 to 187 for modelling or

- assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations
200. Use of a set or device according to any of claims 175 to 187 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
201. Use of a set or device according to any of claims 175 to 187 for predicting optimum configuration/management of thereapeutic intervention.
202. A method according to claim 190 or 191 in which the identification of gene variants is indicative of a higher risk of developing dementia and/or its associated symptoms for the patient or individual.
203. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop dementia and/or its associated symptoms, which method comprises:
- i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from dementia and/or its associated symptoms;
  - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the dementia and/or its associated symptoms;
  - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 175 to 181;
  - iv) calculating the frequencies of these alleles in the samples from i) and ii);
  - v) comparing the frequencies of these alleles in i) and ii);
  - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing dementia and/or its associated symptoms.
204. A method for assessing whether a given subject will be at risk of developing dementia and/or its associated symptoms, which comprises comparing said subject's genotype with a model generated by the method of claim 203.
205. A method according to any of claims 190, 191, 203 and 204 wherein at least one step is computer-controlled.
206. An assay suitable for use in a method according to any of claims 190, 191, 203 and 204; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 175 to 181 in a biological sample.
207. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing dementia and/or its associated symptoms; said kit comprising:
- i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 175 or 177 to 181 in a sample of human DNA;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing dementia and/or its associated symptoms.
208. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing dementia and/or its associated symptoms; said kit comprising:
- i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core



- group of genes as defined in any of claims 176 to 181 in an expressed-protein-containing human sample;
- ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing dementia and/or its associated symptoms.
209. A set of probes according to claim 175, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.
210. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to psychotic disorders and disorders of personality; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

#### KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME  
 T TRANSPORT & STORAGE  
 S STRUCTURAL  
 I IMMUNITY  
 N NERVOUS TRANSMISSION  
 G GROWTH & DIFFERENTIATION

PSYCHOSES & PERSONALITY GENE LIST	HUGO gene symbol	Protein function
11beta hydroxysteroid dehydrogenase 2	HSD11B2	E
5,10-methylenetetrahydrofolate reductase (NADPH)	MTHFR	E
Acetylcholine receptor, nicotinic, alpha A1	CHRNA1	N
Acetylcholine receptor, nicotinic, alpha A2	CHRNA2	N
Acetylcholine receptor, nicotinic, alpha A3	CHRNA3	N
Acetylcholine receptor, nicotinic, alpha A4	CHRNA4	N
Acetylcholine receptor, nicotinic, alpha A5	CHRNA5	N
Acetylcholine receptor, nicotinic, alpha A6	CHRNA6	N
Acetylcholine receptor, nicotinic, alpha A7	CHRNA7	N
Acetylcholine receptor, nicotinic, beta 1	CHRNA1	N
Acetylcholine receptor, nicotinic, beta 2	CHRNA2	N
Acetylcholine receptor, nicotinic, beta 3	CHRNA3	N
Acetylcholine receptor, nicotinic, beta 4	CHRNA4	N
Acetylcholine receptor, nicotinic, epsilon	CHRNA5	N
Acetylcholine receptor, nicotinic, gamma	CHRNA6	N
Acetylcholinesterase	ACHE	E
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N

Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adenylosuccinate lyase	ADSL	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
Adrenocorticotrophic hormone (ACTH) receptor	ACTHR	G
Albumin, ALB	ALB	T
alpha1-antichymotrypsin	AACT	E
alpha-synuclein	SNCA	N
Amyloid beta A4 precursor protein	APP	N
Amyloid beta A4 precursor-like protein	APLP	N
Apolipoprotein A I	APOA1	T
Apolipoprotein A II	APOA2	T
Apolipoprotein B	APOB	T
Apolipoprotein C1	APOC1	T
Apolipoprotein C2	APOC2	T
Apolipoprotein C3	APOC3	T
Apolipoprotein D	APOD	T
Apolipoprotein E	APOE	T
Apolipoprotein H	APOH	T
Arginosuccinate synthetase	ASS	E
Arylsulfatase A	ARSA	E
Ataxia telangiectasia gene, AT	ATM	G
ATP/ADP translocase		E
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G
Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
Bagpipe homeobox, drosophila homolog of, 1	BAPX1	G
beta-synuclein	SNCB	N
Brain derived neurotrophic factor	BDNF	G
Brain derived neurotrophic factor (BDNF) receptor	BDNFR	G
C1 inhibitor		E
Ca(2+) transporting ATPase, slow twitch	ATP2A2	T

Calbindin 1	CALB1	G
Calbindin D9K	CALB3	G
Calcineurin A1	CALNA1	I
Calcineurin A2	CALNA2	I
Calcineurin A3	CALNA3	I
Calcineurin B		I
Calcitonin/Calcitonin gene-related peptide alpha	CALCA	N
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha- 1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha- 1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha- 1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha- 1E (CACNL1A6)	CACNA1E	N
Calcium channel, voltage-dependent, Alpha- 2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N
Calcium channel, voltage-dependent, T-type		N
Calmodulin 1	CALM1	G
Calmodulin 2	CALM2	G
Calmodulin 3	CALM3	G
Calmodulin-dependant protein kinase II	CAMK2A	G
Calnexin	CANX	G
Calpain	CAPN, CAPN3	E
Calretinin	CALB2	N
Cannabinoid receptor	CNR1	N
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Cardiac-specific homeobox, CSX	CSX	G
Caspase 1	CASP1	G
Catechol-O-methyltransferase	COMT	E
Ceroid lipofuscinosis neuronal 2	CLN2	N
Ceroid lipofuscinosis neuronal 3	CLN3	N
Ceroid lipofuscinosis neuronal 4	CLN4	N
Ceroid lipofuscinosis neuronal 5	CLN5	N
Ceroid lipofuscinosis neuronal 6	CLN6	N
Chemokine receptor CCR5	CCR5	I
Chemokine receptor CXCR4	CXCR4	I
Cholecystokinin	CCK	N

Cholecystikinin B receptor	CCKBR	N
Choline acetyltransferase	CHAT	E
Chymotrypsinogen		E
Ciliary neurotrophic factor (CNTF)	CNTF	G
Ciliary neurotrophic factor (CNTF) receptor	CNTFR	G
Citrate synthase		E
Colony-stimulating factor 2	CSF2	G
Colony-stimulating factor 2 alpha receptor	CSF2RA	G
Corticotrophin-releasing hormone	CRH	T
Corticotrophin-releasing hormone receptor	CRHR1	T
Cu <sup>2+</sup> transporting ATPase beta polypeptide	ATP7B	E
Cyclic AMP response element binding protein	CREB	G
Cyclic AMP-dependent protein kinase	PKA	E
Cyclic nucleotide phosphodiesterase 1B	PDE1B	E
Cyclic nucleotide phosphodiesterase 1B1	PDE1B1	E
Cyclic nucleotide phosphodiesterase 2A3	PDE2A3	E
Cyclic nucleotide phosphodiesterase 3A	PDE3A	E
Cyclic nucleotide phosphodiesterase 3B	PDE3B	E
Cyclic nucleotide phosphodiesterase 4A	PDE4A	E
Cyclic nucleotide phosphodiesterase 4C	PDE4C	E
Cyclic nucleotide phosphodiesterase 5A	PDE5A	E
Cyclic nucleotide phosphodiesterase 6A	PDE6A	E
Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E
Cyclic nucleotide phosphodiesterase 8	PDE8	E
Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
CYP11A1	CYP11A1	E
CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E
CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E
CYP2C18	CYP2C18	E
CYP2C19	CYP2C19	E

CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E
CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E
CYP2F1	CYP2F1	E
CYP2J2	CYP2J2	E
CYP3A3	CYP3A3	E
CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystathionase	CTH	E
Cystathione beta synthase	CBS	E
Cytidine deaminase	CDA	E
Cytidine-5-prime-triphosphate synthetase	CTPS	E
Cytochrome a		E
Cytochrome c		E
Cytochrome c oxidase, MTCO		E
Delta aminolevulinate dehydratase	ALAD	E
Delta-7-dehydrocholesterol reductase	DHCR7	E
Dihydrolipoamide succinyltransferase		N
Dopamine beta hydroxylase	DBH	E
Dopamine receptors D1	DRD1	N
Dopamine receptors D2	DRD2	N
Dopamine receptors D3	DRD3	N
Dopamine receptors D4	DRD4	N
Dopamine receptors D5	DRD5	N
Endothelin 1	EDN1	N
Endothelin 2	EDN2	N
Endothelin 3	EDN3	N
Endothelin converting enzyme	ECE1	N
Endothelin receptor type A	EDNRA	N
Endothelin receptor type B	EDNRB	N
Enolase	ENO1	E
Epidermal growth factor	EGF	G
Epidermal growth factor receptor	EGFR	G
Excision repair complementation group 4 protein	ERCC4	E
Fibroblast growth factor	FGF1	G
Fibroblast growth factor receptor 1	FGFR1	G
Fibroblast growth factor receptor 2	FGFR2	G

Fibroblast growth factor receptor 3	FGFR3	G
Flightless-II, Drosophila homolog of	FLII	G
Fragile site, folic acid type, rare, fra(X) A	FRAXA	N
Fragile site, folic acid type, rare, fra(X) E	FRAXE	N
Fragile site, folic acid type, rare, fra(X) F	FRAXF	N
GABA receptor, alpha 1	GABRA1	N
GABA receptor, alpha 2	GABRA2	N
GABA receptor, alpha 3	GABRA3	N
GABA receptor, alpha 4	GABRA4	N
GABA receptor, alpha 5	GABRA5	N
GABA receptor, alpha 6	GABRA6	N
GABA receptor, beta 1	GABRB1	N
GABA receptor, beta 2	GABRB2	N
GABA receptor, beta 3	GABRB3	N
GABA receptor, gamma 1	GABRG1	N
GABA receptor, gamma 2	GABRG2	N
GABA receptor, gamma 3	GABRG3	N
GABA transaminase	ABAT	E
GDP dissociation inhibitor 1	GDI1	G
Geniospasm 1	GSM1	G
Glial-cell derived neurotrophic factor (GDNF) receptor		N
Glial-cell derived neurotrophic factor, GDNF	GDNF	N
Glutamate decarboxylase, GAD	GAD1	E
Glutamate receptor 1	GLUR1	N
Glutamate receptor 2	GLUR2	N
Glutamate receptor 3	GLUR3	N
Glutamate receptor 4	GLUR4	N
Glutamate receptor 5	GLUR5	N
Glutamate receptor 6	GLUR6	N
Glutamate receptor 7	GLUR7	N
Glutamate receptor, ionotropic, NMDA 1	NMDAR1	N
Glutamate receptor, ionotropic, NMDA 2A	NMDAR2A	N
Glutamate receptor, ionotropic, NMDA 2B	NMDAR2B	N
Glutamate receptor, ionotropic, NMDA 2C	NMDAR2C	N
Glutamate receptor, ionotropic, NMDA 2D	NMDAR2D	N
Glutaryl-CoA dehydrogenase	GCDH	E
Glutathione	GSH	T
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
Glycerol kinase	GK	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
Gonadotropin releasing hormone receptor	GNRHR	G
Guanidinoacetate N-methyltransferase	GAMT	E
Guanine nucleotide-binding protein, alpha activating activity polypeptide, GNAO	GNAO1	N

Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 1, GNAI1	GNAI1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 2, GNAI2	GNAI2	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 3, GNAI3	GNAI3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS1	GNAS1	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS2	GNAS2	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS3	GNAS3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS4	GNAS4	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT1	GNAT1	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT2	GNAT2	N
Guanine nucleotide-binding protein, beta polypeptide 3	GNB3	N
Guanine nucleotide-binding protein, q polypeptide	GNAQ	N
Guanylate cyclase 2D, membrane (retina-specific)	GUCY2D	E
Guanylate cyclase activator 1A (retina)	GUCA1A	E
Guanylyl cyclase		E
Heat shock protein, HSP60		I
Heat shock protein, HSP70		I
Heat shock protein, HSP90		I
Heat shock protein, HSPA1		I
Heat shock protein, HSPA2		I
Heparan sulfamidase		E
Hepatic lipase	LIPC	E
Histamine receptors, H1		N
Histamine receptors, H2		N
Histamine receptors, H3		N
HMG-CoA reductase	HMGCR	E
Huntingtin	HD	T
Hypoxanthine-guanine phosphoribosyltransferase, HGPRT	HPRT	E
Hypoxia inducible factor 1	HIF1A	E
Hypoxia inducible factor 2		E
Inositol monophosphatase	IMPA1	N
Insulin	INS	G
Insulin receptor	INSR	G
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I

Interleukin(IL) 10	IL10	I
Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I
Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
IP3 kinase		E
Leukin		I
Mismatch repair gene, PMSL2	PMS2	G
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Myelin basic protein		S
Myosin, light chain 3	MYL3	S
NADPH-dependent cytochrome P450 reductase	POR	E
Nerve growth factor	NGF	G
Nerve growth factor receptor	NGFR	G
Neurite inhibitory protein		N
Neurofibromin 1	NF1	G
Neurofibromin 2	NF2	G
Neurofilament protein, NF125	NF150	S
Neurofilament protein, NF200	NF200	S
Neurofilament protein, NF68	NF68	S



Neurokinin A	NKNA	N
Neurokinin B	NKNB	N
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Neurotensin	NTS	N
Neurotensin receptor	NTSR1	N
Nitric oxide synthase 1, NOS1	NOS1	E
Nitric oxide synthase 2, NOS2	NOS2	E
Nitric oxide synthase 3, NOS3	NOS3	E
Nucleoside diphosphate kinase-A	NDPKA	E
Oncogene sis	PDGFB	G
Opioid receptor, delta	OPRD1	N
Opioid receptor, kappa	OPRK1	N
Opioid receptor, mu	OPRM1	N
Ornithine delta-aminotransferase	OAT	E
Paraoxonase PON1	PON1	E
Parkin	PARK2	N
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C alpha		I
Phospholipase C beta		I
Phospholipase C delta	PLCD1	I
Phospholipase C epsilon		I
Phospholipase C gamma	PLCG1	I
Platelet derived growth factor	PDGF	G
Platelet derived growth factor receptor	PDGFR	G
Potassium inwardly-rectifying channel J1	KCNJ1	N
POU domain, class 1, transcription factor 1 (Pit1)	POU1F1	G
Presenilin 1	PSEN1	T
Presenilin 2	PSEN2	T
Prion protein	PRNP	N
Proline dehydrogenase	PRODH	E
Proopiomelanocortin	POMC	N
Prosaposin	PSAP	N
Protective protein for beta-galactosidase	PPGB	E
Protein kinase C, alpha	PRKCA	E
Protein kinase C, gamma	PRKCG	E
Protein kinase G		E
Protein phosphatase 1, regulatory (inhibitor) subunit 3	PPP1R3	E

Proteolipid protein	PLP	N
RIGUI	RIGUI	G
S100 calcium-binding protein A1	S100A1	N
S100 calcium-binding protein A2	S100A2	N
S100 calcium-binding protein A3	S100A3	N
S100 calcium-binding protein A4	S100A4	N
S100 calcium-binding protein A5	S100A5	N
S100 calcium-binding protein A6	S100A6	N
S100 calcium-binding protein A7	S100A7	N
S100 calcium-binding protein A8	S100A8	N
S100 calcium-binding protein A9	S100A9	N
S100 calcium-binding protein B	S100B	N
S100 calcium-binding protein P	S100P	N
Secretase, alpha		N
Secretase, beta		N
Secretase, gamma		N
Serotonin N-acetyltransferase	SNAT	E
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Solute carrier family 12, member 1	SLC12A1	T
Solute carrier family 12, member 2	SLC12A2	T
Solute carrier family 12, member 3	SLC12A3	T
Solute carrier family 4 (anion exchanger), member 1	SLC4A1	T
Solute carrier family 4 (anion exchanger), member 2	SLC4A2	T
Solute carrier family 4 (anion exchanger), member 3	SLC4A3	T
Solute carrier family 5 (sodium/glucose transporter), member 1	SLC5A1	T
Solute carrier family 5 (sodium/glucose	SLC5A2	T

transporter), member 2		
Solute carrier family 5 (sodium/glucose transporter), member 5	SLC5A5	T
Solute carrier family 5, member 3	SLC5A3	T
Solute carrier family 6 (GAMMA-AMINO-BUTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Solute carrier family 6 (neurotransmitter transporter, serotonin), member 4	SLC6A4	T
Superoxide dismutase 1	SOD1	E
Superoxide dismutase 3	SOD3	E
Synapsin 1a & 1b	SYN1	N
Synapsin 2a & 2b	SYN2	N
Synaptic vesicle amine transporter	SVAT	N
Synaptogyrin		N
Synaptophysin	SYP	N
Synaptosomal-associated protein, 25KD	SNAP25	N
Syntaxin 1	STX1	N
Tachykinin receptor, NK1R	TACR1	N
Tachykinin receptor, NK2R	TACR2	N
Tachykinin receptor, NK3R	TACR3	N
Talin	TLN	G
TEK, tyrosine kinase, endothelial	TEK	E
Telomerase protein component		E
Transcobalamin 1, TCN1		T
Transcobalamin 2, TCN2	TCN2	T
Transcription factor, TUPLE1	TUPLE1	N
Transforming growth factor, beta 2	TGFB2	G
Transforming growth factor, beta induced	TGFB1	G
Transforming growth factor, beta receptor 2	TGFBR2	G
Transthyretin	TTR	T
Trypsin inhibitor		E
Tryptophan 2,3-dioxygenase	TDO2	N
Tryptophan hydroxylase	TPH	E
Tumour necrosis factor (TNF) receptor associated factor 1	TRAF1	I
Tumour necrosis factor (TNF) receptor associated factor 2	TRAF2	I
Tumour necrosis factor (TNF) receptor associated factor 3	TRAF3	I
Tumour necrosis factor (TNF) receptor associated factor 4	TRAF4	I
Tumour necrosis factor (TNF) receptor associated factor 5	TRAF5	I
Tumour necrosis factor (TNF) receptor	TRAF6	I

associated factor 6		
Tumour necrosis factor alpha	TNFA	I
Tumour necrosis factor alpha receptor	TNFAR	I
Tumour necrosis factor beta	TNFB	I
Tumour necrosis factor beta receptor	TNFBR	I
Tyrosinase	TYR	E
Tyrosine hydroxylase	TH	E
Ubiquitin		G
Ubiquitin activating enzyme, E1		E
Ubiquitin B	UBB	G
Ubiquitin C	UBC	G
Ubiquitin protein ligase E3A	UBE3A	E
UDP-glucuronosyltransferase 1	ugt1d, UGT1	E
UDP-glucuronosyltransferase 2	UGT2	E
Uridinediphosphate(UDP)-galactose-4-epimerase	GALE	E
Vacuolar proton pump, subunit 1	VPP1	N
Vacuolar proton pump, subunit 3	VPP3	N
Vesicular monoamine transporter 1	VMAT1	N
Vesicular monoamine transporter 2	VMAT2	N

211. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 210.

212. A set according to claim 210 or 211 in which a minority of said probes for listed genes are absent.

213. A set according to claim 210 or 211 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.

214. A set according to claim 210 or 211 in which a limited number of probes are replaced by probes for non-listed genes.

215. A set of probes for a core group of genes according to any of claims 210 to 214 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.

216. A set according to any of claims 210 to 215 consisting of probes for members of a sub-group of the core group.

217. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.

218. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.
219. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.
220. A set according to claim 217 or 218 in which said substrate is a semiconductor microchip.
221. A set according to any preceding claim for use in a biological assay for detection of said gene variants.
222. A set according to any preceding claim for use in the measurement of differential gene expression levels.
223. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
224. A medical device including a set according to any of claims 210 to 222 for use in an array for detection of differential gene expression levels.
225. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 210) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 210 and 212 to 222 and relating the probe hybridisation pattern to said variations.
226. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 211) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 211 to 222 and relating the probe interaction pattern to said variations.
227. Use of a set or device according to any of claims 210 to 222 for the prognosis and management of patients suffering from or at risk of experiencing the symptoms and consequences of psychotic disorders and disorders of personality.
228. Use of a set or device according to any of claims 210 to 222 for predicting likely therapeutic response and adverse events following therapeutic intervention.
229. Use of a set or device according to any of claims 210 to 222 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
230. Use of a set or device according to any of claims 210 to 222 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
231. Use of a set or device according to any of claims 210 to 222 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
232. Use of a set or device according to any of claims 210 to 222 for the development of new strategies of therapeutic intervention and in clinical trials.
233. Use of a set or device according to any of claims 210 to 222 for construction of and generation of algorithms for patient and healthcare management.
234. Use of a set or device according to any of claims 210 to 222 for modelling or assessing the impact of diseases or healthcare management strategies on

- individuals, groups, patient cohorts or populations
235. Use of a set or device according to any of claims 210 to 222 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
236. Use of a set or device according to any of claims 210 to 222 for predicting optimum configuration/management of thereapeutic intervention.
237. A method according to claim 225 or 226 in which the identification of gene variants is indicative of a higher risk of developing the symptoms and consequences of psychotic disorders and disorders of personality for the patient or individual.
238. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop the symptoms and consequences of psychotic disorders and disorders of personality, which method comprises:
- i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from the symptoms and consequences of psychotic disorders and disorders of personality;
  - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the symptoms and consequences of psychotic disorders and disorders of personality;
  - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 210 to 216;
  - iv) calculating the frequencies of these alleles in the samples from i) and ii);
  - v) comparing the frequencies of these alleles in i) and ii);
  - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing the symptoms and consequences of psychotic disorders and disorders of personality.
239. A method for assessing whether a given subject will be at risk of developing the symptoms and consequences of psychotic disorders and disorders of personality, which comprises comparing said subject's genotype with a model generated by the method of claim 238.
240. A method according to any of claims 225, 226, 238 and 239 wherein at least one step is computer-controlled.
241. An assay suitable for use in a method according to any of claims 225, 226, 238 and 239; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 210 to 216 in a biological sample.
242. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing the symptoms and consequences of psychotic disorders and disorders of personality; said kit comprising:
- i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 210 or 210 to 216 in a sample of human DNA;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing the symptoms and consequences of psychotic disorders and disorders of personality.

243. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing the symptoms and consequences of psychotic disorders and disorders of personality; said kit comprising:
- i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core group of genes as defined in any of claims 211 to 216 in an expressed-protein-containing human sample;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing the symptoms and consequences of psychotic disorders and disorders of personality.
244. A set of probes according to claim 210, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.
245. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to cardiovascular disease, dysfunction and/or damage; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

#### KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME  
 T TRANSPORT & STORAGE  
 S STRUCTURAL  
 I IMMUNITY  
 N NERVOUS TRANSMISSION  
 G GROWTH & DIFFERENTIATION

CARDIOVASCULAR GENE LIST	HUGO gene symbol	Protein function
17beta hydroxysteroid oxidoreductase		E
2,3-bisphosphoglycerate mutase	BPGM	E
3 beta hydroxysteroid dehydrogenase 2	HSD3B2	E
3-oxoacid CoA transferase	OXCT	E
5,10-methylenetetrahydrofolate reductase (NADPH)	MTHFR	E
Acetoacetyl 1-CoA-thiolase	ACAT1	E
Acetoacetyl 2-CoA-thiolase	ACAT2	E
Acetyl CoA acyltransferase	ACAA	E
Acetylcholinesterase	ACHE	E
Acid phosphatase 2, lysosomal	ACP2	E
Acidic amino acid transporter		T
Actin, alpha, cardiac	ACTC	S
Actin, alpha, skeletal	ACTA1	S

Actin, alpha, smooth, aortic	ACTA2	S
Activin A receptor, type 2B	ACVR2B	G
Acyl CoA dehydrogenase, long chain	ACADL	E
Acyl CoA dehydrogenase, very long chain	ACADVL	E
Adaptin, beta 3A	ADTB3A	T
Adducin, alpha	ADD1	S
Adducin, beta	ADD2	S
Adenosine deaminase	ADA	E
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N
Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adenylate kinase	AK1	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
Adrenocorticotrophic hormone (ACTH) receptor	ACTHR	G
Alanine aminotransferase		T
Alanine-glyoxylate aminotransferase	AGXT	E
Albumin, ALB	ALB	T
Alcohol dehydrogenase 1	ADH1	E
Alcohol dehydrogenase 2	ADH2	E
Alcohol dehydrogenase 3	ADH3	E
Alcohol dehydrogenase 5	ADH5	E
Alcohol dehydrogenase 6	ADH6	E
Alcohol dehydrogenase 7	ADH7	E
Aldehyde dehydrogenase 1	ALDH1	E
Aldehyde dehydrogenase 10	ALDH10	E
Aldehyde dehydrogenase 2	ALDH2	E
Aldehyde dehydrogenase 5	ALDH5	E
Aldehyde dehydrogenase 6	ALDH6	E
Aldehyde dehydrogenase 7	ALDH7	E
Aldolase A	ALDOA	E
Aldolase B	ALDOB	E
Aldolase C	ALDOC	E
Aldosterone receptor	MLR	G



Alpha 1 acid glycoprotein	AAG; AGP	T
Alpha 2 macroglobulin	A2M	I
alpha1-antitrypsin	PI	E
alpha2-antiplasmin	PLI	E
alpha-actinin 2	ACTN2	G
alpha-actinin 3	ACTN3	G
alpha-Galactosidase A	GLA	E
alpha-L-Iduronidase	IDUA	E
Aminopeptidase P	XPNPEP2	E
Amphiregulin	AREG	G
Amylo-1,6-glucosidase	AGL	E
Angiopoietin 1	ANGPT1	G
Angiopoietin 2	ANGPT2	G
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensin receptor 1	AGTR1	T
Angiotensin receptor 2	AGTR2	T
Angiotensinogen	AGT	E
Ankyrin 1	ANK1	S
Ankyrin 2	ANK2	S
Ankyrin 3	ANK3	S
Annexin 1	ANX 1	I
Antidiuretic hormone receptor	ADHR	T
Antithrombin III	AT3	E
Apolipoprotein (a)	LPA	T
Apolipoprotein A 4	APOA4	T
Apolipoprotein A I	APOA1	T
Apolipoprotein A II	APOA2	T
Apolipoprotein B	APOB	T
Apolipoprotein C1	APOC1	T
Apolipoprotein C2	APOC2	T
Apolipoprotein C3	APOC3	T
Apolipoprotein D	APOD	T
Apolipoprotein E	APOE	T
Apolipoprotein H	APOH	T
Aquaporin 1	AQP1	T
Aquaporin 2	AQP2	T
Arginine vasopressin	AVP	N
Arginine vasopressin receptor 1A	AVPR1A	N
Arginine vasopressin receptor 1B	AVPR1B	N
Arginine vasopressin receptor 2	AVPR2	N
Arginosuccinate lyase	ASL	E
Arylsulfatase B	ARSB	E
Aspartylglucosaminidase	AGA	E
Ataxia telangiectasia gene, AT	ATM	G
ATP/ADP translocase		E
ATP-binding cassette transporter 7	ABC7	I
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G

Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
Autoimmune regulator, AIRE	AIRE	I
BCL2-related protein A1	BCL2A1	G
beta 2 microglobulin	B2M	I
beta-endorphin receptor		N
Bile acid coenzyme A: amino acid N-acyltransferase	BAAT	E
Bile salt export pump	BSEP, PFIC2	T
Bile salt-stimulated lipase	CEL	E
Bilirubin UDP-glucuronosyltransferase		E
Bloom syndrome protein	BLM	G
Bradykinin receptor B1		I
Bradykinin receptor B2		I
Butyrylcholinesterase	BCHE	E
Ca(2+) transporting ATPase, fast twitch	ATP2A1	T
Ca(2+) transporting ATPase, slow twitch	ATP2A2	T
Cadherin E	CDH1	G
Cadherin EP		G
Cadherin N	CDH2	G
Cadherin P	CDH3	G
Calbindin 1	CALB1	G
Calbindin D9K	CALB3	G
Calcineurin A1	CALNA1	I
Calcineurin A2	CALNA2	I
Calcineurin A3	CALNA3	I
Calcineurin B		I
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha-1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha-1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha-1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha-1E (CACNL1A6)	CACNA1E	N
Calcium channel, voltage-dependent, Alpha-2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, L type, alpha 1S subunit	CACNA1S	N
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N
Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	CACNA1A	N
Calcium channel, voltage-dependent, T-type		N

Hepatic lipase	LIPC	E
Hermansky-pudlak syndrome gene	HPS	T
Hexokinase 1	HK1	E
Hexosaminidase A	HEXA,TSD	E
Hexosaminidase B	HEXB	E
Histidine-rich glycoprotein	HRG	T
HLA-B associated transcript 1	BAT1	I
HLH transcription factor HAND1	HAND1	G
HLH transcription factor HAND2	HAND2	G
HMG-CoA lyase	HMGCL	E
HMG-CoA reductase	HMGCR	E
HMG-CoA synthase	HMGCS2	E
Homeobox (HOX) gene A13	HOXA13	G
Homeobox HB24	HLX1	G
Hormone-sensitive lipase	HSL	E
Human chorionic gonadotrophin, hCG	CG	G
Human placental lactogen	CSH1	G
Hypoxanthine-guanine phosphoribosyltransferase, HGPRT	HPRT	E
Hypoxia inducible factor 1	HIF1A	E
Hypoxia inducible factor 2		E
IC7 A and B		I
Iduronate 2 sulphatase	IDS	E
Indian hedgehog, ihh	IHH	G
Inosine triphosphatase	ITPA	E
Inositol 1,4,5-triphosphate receptor 1	ITPR1	G
Inositol 1,4,5-triphosphate receptor 3	ITPR3	G
Inositol monophosphatase	IMPA1	N
Inositol polyphosphate 1-phosphatase	INPP1	N
Insulin	INS	G
Insulin receptor	INSR	G
Insulin receptor substrate-1	IRS1	G
Insulin-like growth factor 1	IGF1	G
Insulin-like growth factor 1 receptor	IGF1R	G
Insulin-like growth factor 2	IGF2	G
Insulin-like growth factor 2 receptor	IGF2R	G
Integrin beta 1	ITGB1	G
Integrin beta 2	ITGB2	G
Integrin beta 3	ITGB3	G
Integrin beta 4	ITGB4	G
Integrin beta 5	ITGB5	G
Integrin beta 6	ITGB6	G
Integrin beta 7	ITGB7	G
Integrin, alpha 1	ITGA1	G
Integrin, alpha 2	ITGA2	G
Integrin, alpha 3	ITGA3	G
Integrin, alpha 4	ITGA4	G
Integrin, alpha 5	ITGA5	G

Integrin, alpha 6	ITGA6	G
Integrin, alpha 7	ITGA7	G
Integrin, alpha 8	ITGA8	G
Integrin, alpha 9	ITGA9	G
Integrin, alpha M	ITGAM	G
Integrin, alpha X	ITGAX	G
Inter-alpha-trypsin inhibitor, IAT		E
Intercellular adhesion molecule 1	ICAM1	I
Intercellular adhesion molecule 2	ICAM2	I
Intercellular adhesion molecule 3	ICAM3	I
Interferon alpha	IFNA1	I
Interferon beta	IFNB	I
Interferon gamma	IFNG	I
Interferon gamma receptor 1	IFNGR1	I
Interferon gamma receptor 2	IFNGR2	I
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) 10	IL10	I
Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I
Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
IP3 kinase		E
Isovaleric acid CoA dehydrogenase	IVD	E
Kallikrein 3	KAK3	I
Kell blood group precursor	XK, KEL	T

Ketohexokinase	KHK	E
Kininogen, High molecular weight	KNG	I
Kynureninase		E
Lactate dehydrogenase, A	LDHA	E
Lactate dehydrogenase, B	LDHB	E
Lamin A/C	LMNA	G
Laminin 5, alpha 3	LAMA3	G
Laminin 5, beta 3	LAMB3	G
Laminin 5, gamma 2	LAMC2	G
Laminin M	LAMM	G
Laminin receptor 1	LAMR1	G
Latent transforming growth factor-beta binding protein 2	LTBP2	G
Lecithin-cholesterol acyltransferase	LCAT	E
Lectin, mannose-binding 1	LMAN1	I
Lectin, mannose-binding 2	MBL2	I
Leptin	LEP	G
Leptin receptor	LEPR	G
Leukocyte-specific transcript 1	LST-1	I
Leukotriene A4 synthase	LTA4S	E
Leukotriene B4 receptor		I
Leukotriene B4 synthase	LTB4S	E
Leukotriene C4 synthase	LTC4S	E
LIM homeobox protein 1	LHX1	G
Lipocortin 1	ANX4	I
Lipoprotein lipase	LPL	I
Lipoprotein receptor, Low Density	LDLR	T
Lipoprotein, High Density	HDLDT1	T
Lipoprotein, Very Low Density	VLDLR	T
Lipoprotein-associated coagulation factor	LACI	I
Lipoxygenase		E
Lipoxygenase 12 (platelets)	LOG12	I
Long QT-type 2 potassium channels	LQT2, KCNH2	T
Low density lipoprotein receptor-related protein precursor	LRP	T
Lymphoid enhancer-binding factor	LEF-1	G
Lysosomal acid lipase	LIPA	E
Macrophage inflammatory protein-2	MIP2	I
MAD (mothers against decapentaplegic, Drosophila) homologue 4	MADH4	G
MADS box transcription-enhancer factor 2A	MEF2A	G
MADS box transcription-enhancer factor 2B	MEF2B	G
Mannosidase, alpha B lysosomal	MANB	E
Matrix Gla protein	MGP	G
Matrix metalloproteinase 1	MMP1	E
Matrix metalloproteinase 10	MMP10	E
Matrix metalloproteinase 11	MMP11	E
Matrix metalloproteinase 12	MMP12	E

Matrix metalloproteinase 13	MMP13	E
Matrix metalloproteinase 14	MMP14	E
Matrix metalloproteinase 15	MMP15	E
Matrix metalloproteinase 16	MMP16	E
Matrix metalloproteinase 17	MMP17	E
Matrix metalloproteinase 18	MMP18	E
Matrix metalloproteinase 19	MMP19	E
Matrix metalloproteinase 2	MMP2	E
Matrix metalloproteinase 3	MMP3, STMY1	E
Matrix metalloproteinase 4	MMP4	E
Matrix metalloproteinase 5	MMP5	E
Matrix metalloproteinase 6	MMP6	E
Matrix metalloproteinase 7	MMP7	E
Matrix metalloproteinase 8	MMP8	E
Matrix metalloproteinase 9	MMP9	E
Melanocortin 2 receptor	MC2R	T
Melanocortin 4 receptor	MC4R	T
Methionine synthase	MTR	E
Methionine synthase reductase	MTRR	E
Methylmalonyl-CoA mutase	MUT	E
Mevalonate kinase	MVK	E
MHC Class I: A		I
MHC Class I: B		I
MHC Class I: C		I
MHC Class I: LMP-2, LMP-7		I
MHC Class I: Tap1	ABCR, TAP1	I
MHC Class II: DP	HLA-DPB1	I
MHC Class II: DQ		I
MHC Class II: DR		I
MHC Class II: Tap2	TAP2, PSF2	I
MHC Class II:Complementation group A	MHC2TA	I
MHC Class II:Complementation group B	rfxank	I
MHC Class II:Complementation group C	RFX5	I
MHC Class II:Complementation group D	RFXAP	I
Microsomal triglyceride transfer protein	MTP	T
Mismatch repair gene, PMSL2	PMS2	G
Mitochondrial trifunctional protein, alpha subunit	HADHA	E
Mitochondrial trifunctional protein, beta subunit	HADHB	E
Molybdenum cofactor synthesis 1	MOCS1	E
Molybdenum cofactor synthesis 2	MOCS2	E
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Monocyte chemoattractant protein 1	MCP1	I
Mucopolidoses	GNPTA	E
Mulibrey nanism	MUL	T
Muscarinic receptor, M1	CHRM1	N

Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
MutS homolog 3	MSH3	G
Myoglobin		T
Myosin, cardiac	MYH7	S
Myosin, light chain 2	MYL2	S
Myosin, light chain 3	MYL3	S
Myosin-binding protein C, cardiac	MYBPC3	S
Myotubularin	MTM1	S
Na <sup>+</sup> , K <sup>+</sup> ATPase, alpha	ATP1A1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 1	ATP1B1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 2	ATP1B2	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 3	ATP1B3	G
Na <sup>+</sup> /H <sup>+</sup> exchanger 1	NHE1	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 2	NHE2	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 3	NHE3	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 4	NHE4	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 5	NHE5	T
N-acetylglucosamine-6-sulfatase	GNS	E
NADPH oxidase		I
NADPH-dependent cytochrome P450 reductase	POR	E
NB6		I
Nebulin	NEB	S
Nephronophthisis 1	NPHP1	T
Neuraminidase sialidase	NEU	T
Neuregulin	HGL	G
Neurite inhibitory protein		N
Neuroendocrine convertase 1	NEC1, PCSK1	E
Neurokinin A	NKNA	N
Neurokinin B	NKNB	N
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Neutrophil cystolic factor 1	NCF1	I
Neutrophil cystolic factor 2	NCF2	I
Niemann-Pick disease protein	NPC1	T
Nitric oxide synthase 1, NOS1	NOS1	E
Nitric oxide synthase 2, NOS2	NOS2	E
Nitric oxide synthase 3, NOS3	NOS3	E
Notch ligand - jagged 1	JAG1, AGS	G
Nuclear factor I-kappa-B-like gene	IKBL	I
Oncogene sis	PDGFB	G
Oncostatin M	OSM	G
Oncostatin M receptor	OSMR	G
Osteonectin	ON	G

Osteopontin	OPN	G
Osteoprotegerin	OPG	G
Pancreatic lipase	PNLIP	E
Pancreatic lipase related protein 1	PLRP1	E
Pancreatic lipase related protein 2	PLRP2	E
Paraoxonase PON1	PON1	E
Paraoxonase PON2	PON2	E
Paraoxonase PON3		E
Parvalbumin	PVALB	G
Patched (Drosophila) homolog, PTCH	PTCH	G
PCNA (proliferating cell nuclear antigen)		E
Pepsinogen		E
Peroxidase, salivary	SAPX	E
Peroxisomal membrane protein 1	PXMP1	S
Peroxisomal membrane protein 3	PXMP3	T
Peroxisome biogenesis factor 1	PEX1	T
Peroxisome biogenesis factor 19	PEX19	T
Peroxisome biogenesis factor 6	PEX6	T
Peroxisome biogenesis factor 7	PEX7	T
Peroxisome proliferative activated receptor, alpha	PPARA	T
Peroxisome proliferative activated receptor, gamma	PPARG	T
Peroxisome receptor 1	PXR1	T
P-glycoprotein 3	PGY3	T
Phosphatidylinositol glycan, class A (paroxysmal nocturnal hemoglobinuria)	PIGA	G
Phosphatidylinositol transfer protein	PITPN	G
Phosphofructokinase, muscle	PFKM	E
Phosphoglucose isomerase	GPI	E
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C alpha		I
Phospholipase C beta		I
Phospholipase C delta	PLCD1	I
Phospholipase C epsilon		I
Phospholipase C gamma	PLCG1	I
Phosphomannomutase-2	PMM2	T
Phosphoribosyl pyrophosphate synthetase	PRPS1	E
Phosphorylase kinase, alpha 2	PHKA2	E
Phytanoyl-CoA hydroxylase	PHYH	G
Plasminogen	PLG	E



Plasminogen activator inhibitor 1	PAI1	E
Plasminogen activator inhibitor 2	PAI2	E
Plasminogen activator receptor, Urokinase	UPAR; PLAUR	S
Plasminogen activator, Tissue	PLAT; TPA	E
Plasminogen activator, Urokinase	UPA; PLAU	E
Platelet derived growth factor	PDGF	G
Platelet derived growth factor receptor	PDGFR	G
Platelet glutaminase	GLS	T
Platelet glycoprotein 1b, alpha	GP1BA	I
Platelet glycoprotein 1b, beta	GP1BB	I
Platelet glycoprotein 1b, gamma	GP1BG	I
Platelet glycoprotein IX	GP9	I
Platelet glycoprotein V	GP5	I
Platelet monamine oxidase		T
Platelet-activating factor acetylhydrolase 1B	PAFAH1B1 or LIS1	I
Platelet-activating factor acetylhydrolase 2	PAFAH2	I
Platelet-activating factor receptor	PAFR	I
Poly (ADP-ribose) synthetase	PARS	E
Polycystic kidney and hepatic disease 1	PKHD1	T
Polycystin 1	PKD1	T
Polycystin 2	PKD2	T
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium inwardly-rectifying channel J11	KCNJ11	N
Potassium voltage-gated channel A1	KCNA1	N
Potassium voltage-gated channel E1	KCNE1	N
Potassium voltage-gated channel Q1	KCNQ1	N
Potassium voltage-gated channel Q2	KCNQ2	N
Potassium voltage-gated channel Q3	KCNQ3	N
POU domain, class 1, transcription factor 1 (Pit1)	POU1F1	G
Prekallikrein		I
Procollagen N-protease		E
Progesterone receptor (RU486 binding receptor)	PGR	G
Pro-melanin-concentrating hormone	PMCH	G
Proopiomelanocortin	POMC	N
Prostaglandin (PG) D synthase, hematopoietic	PGDS	E
Prostaglandin E2 receptor		I
Prostaglandin-endoperoxidase synthase 2	PTGS2	G
Protease inhibitor 1		T
Protease nexin 2	PN2	E
Protective protein for beta-galactosidase	PPGB	E
Protein C	PROC	I
Protein C inhibitor	PCI	I
Protein S	PROS1	I
Prothrombin precursor	F2	I

Protoporphyrinogen oxidase	PPOX	E
Purine nucleoside phosphorylase	NP	E
Purinergic receptor P1A1		N
Purinergic receptor P1A2		N
Purinergic receptor P1A3		N
Purinergic receptor P2X, 1	P2RX1	N
Purinergic receptor P2X, 2	P2RX2	N
Purinergic receptor P2X, 3	P2RX3	N
Purinergic receptor P2X, 4	P2RX4	N
Purinergic receptor P2X, 5	P2RX5	N
Purinergic receptor P2X, 6	P2RX6	N
Purinergic receptor P2X, 7	P2RX7	N
Purinergic receptor P2Y, 1	P2RY1	N
Purinergic receptor P2Y, 11	P2RY11	N
Purinergic receptor P2Y, 2	P2RY2	N
Pyruvate carboxylase	PC	E
Pyruvate decarboxylase	PDHA	E
Pyruvate kinase	PKLR	E
Radixin	RDX	S
Renin	REN	E
Replication factor C	RFC2	E
Retinoic acid receptor, alpha	RARA	G
Retinoic acid receptor, beta	RARB	G
Retinoic acid receptor, gamma	RARG	G
Retinoid X receptor, alpha	RXRA	G
Retinoid X receptor, beta	RXRB	G
Retinoid X receptor, gamma	RXRG	G
Rhesus blood group, CcEe antigens	RHCE	T
Rhesus blood group, D antigen	RHD	T
Rhesus blood group-associated glycoprotein	RHAG	T
Ribosomal protein S19	RPS19	E
RIGUI	RIGUI	G
S100 calcium-binding protein A1	S100A1	N
S100 calcium-binding protein A2	S100A2	N
S100 calcium-binding protein A3	S100A3	N
S100 calcium-binding protein A4	S100A4	N
S100 calcium-binding protein A5	S100A5	N
S100 calcium-binding protein A6	S100A6	N
S100 calcium-binding protein A7	S100A7	N
S100 calcium-binding protein A8	S100A8	N
S100 calcium-binding protein A9	S100A9	N
S100 calcium-binding protein B	S100B	N
S100 calcium-binding protein P	S100P	N
SA homolog	SAH	G
SAP (SLAM-associated protein)	SH2D1A	I
Secretase, alpha		N
Secretase, beta		N
Secretase, gamma		N

Selectin E	SELE	N
Selectin L	SELL	N
Selectin P	SELP	N
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Serum amyloid A	SAA	T
Serum amyloid P	SAP	T
Sjogren (Sjogren) syndrome antigen A1	SSA1	I
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage gated, type IV, alpha polypeptide	SCN4A	N
Sodium channel, voltage gated, type V, alpha polypeptide	SCN5A	N
Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Solute carrier family 1 (glutamate transporter), member 1	SLC1A1	T
Solute carrier family 1 (glutamate transporter), member 2	SLC1A2	T
Solute carrier family 10 (sodium/bile acid cotransporter family), member 1	SLC10A1	T
Solute carrier family 10 (sodium/bile acid cotransporter family), member 2	SLC10A2	T
Solute carrier family 12, member 1	SLC12A1	T
Solute carrier family 12, member 2	SLC12A2	T
Solute carrier family 12, member 3	SLC12A3	T
Solute carrier family 2 (facilitated glucose transporter), member 1	SLC2A1	T
Solute carrier family 2 (facilitated glucose transporter), member 2	SLC2A2	T
Solute carrier family 2 (facilitated glucose transporter), member 3	SLC2A3	T
Solute carrier family 2 (facilitated glucose transporter), member 4	SLC2A4	T

transporter), member 4		
Solute carrier family 2 (facilitated glucose transporter), member 5	SLC2A5	T
Solute carrier family 21, member 2	SLC21A2	T
Solute carrier family 21, member 3	SLC21A3	T
Solute carrier family 22, member 5	SLC22A5	T
Solute carrier family 3 (facilitated glucose transporter), member 1	SLC3A1	T
Solute carrier family 4 (anion exchanger), member 1	SLC4A1	T
Solute carrier family 4 (anion exchanger), member 2	SLC4A2	T
Solute carrier family 4 (anion exchanger), member 3	SLC4A3	T
Solute carrier family 5 (sodium/glucose transporter), member 1	SLC5A1	T
Solute carrier family 5 (sodium/glucose transporter), member 2	SLC5A2	T
Solute carrier family 5 (sodium/glucose transporter), member 5	SLC5A5	T
Solute carrier family 5, member 3	SLC5A3	T
Solute carrier family 6 (GAMMA-AMINOBUTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Solute carrier family 6 (neurotransmitter transporter, serotonin), member 4	SLC6A4	T
Solute carrier family 8 (sodium/calcium exchanger), member 1	SLC8A1	T
Sonic hedgehog, SHH	SHH	G
Sorcin	SRI	T
Spectrin alpha	SPTA1	S
Spectrin beta	SPTB	S
Sphingomyelinase	SMPD1	E
Stem cell factor	SCF	G
Steroid 5 alpha reductase 1	SRD5A1	E
Steroid 5 alpha reductase 2	SRD5A2	E
Steroidogenic acute regulatory protein	STAR	T
Sterol carrier protein 2	SCP2	T
Succinate dehydrogenase 1	SDH1	E
Succinate dehydrogenase 2	SDH2	E
Succinate thiokinase		E
Superoxide dismutase 1	SOD1	E
Superoxide dismutase 3	SOD3	E
Surfeit 1	SURF1	G

Synapsin 1a & 1b	SYN1	N
Synapsin 2a & 2b	SYN2	N
Synaptic vesicle amine transporter	SVAT	N
Synaptobrevin 1	SYB1	N
Synaptobrevin 2	SYB2	N
Synaptogyrin		N
Synaptophysin	SYP	N
Synaptosomal-associated protein, 25KD	SNAP25	N
Synaptotagmin 1	SYT1	N
Synaptotagmin 2	SYT2	N
Syntaxin 1	STX1	N
Talin	TLN	G
T-BOX 1	TBX1	G
T-BOX 3	TBX3	G
TEK, tyrosine kinase, endothelial	TEK	E
Terminal deoxynucleotidyltransferase	TDT	I
Tetranectin	TNA	T
Thiolase, peroxisomal		E
Thiopurine S-methyltransferase	TPMT	E
Thrombin receptor	F2R	I
Thrombomodulin	THBD	I
Thrombopoietin	THPO	G
Thrombospondin	THBS1	G
Thromboxane A synthase 1	TBXAS1	I
Thromboxane A2	TXA2	I
Thromboxane A2 receptor	TBXA2R	I
Thy-1 T-cell antigen	THY1	I
Thymic humoral factor		I
Thymopoietin	TMPO	G
Thymosin		I
Thyroid hormone receptor, alpha	THRA	G
Thyroid hormone receptor, beta	THRB	G
TIE receptor tyrosine kinase	TIE-1	G
Tip-associated protein	TAP	I
Tissue inhibitor of metalloproteinase 1, TIMP1	TIMP1	E
Tissue inhibitor of metalloproteinase 2, TIMP2	TIMP2	E
Tissue inhibitor of metalloproteinase 3, TIMP3	TIMP3	E
Tissue inhibitor of metalloproteinase 4, TIMP4	TIMP4	E
Topoisomerase I		E
Torticollis, keloids, cryptorchidism and renal dysplasia gene	TKCR	G
Transcobalamin 2, TCN2	TCN2	T
Transcription factor 2, hepatic	TCF2	G
Transferrin	TF	G

Transferrin receptor	TFRC	G
Transforming growth factor, beta 2	TGFB2	G
Transforming growth factor, beta induced	TGFB1	G
Transforming growth factor, beta receptor 2	TGFBR2	G
Translocation in renal carcinoma on chromosome 8 gene	TRC8	G
Transthyretin	TTR	T
Triosephosphate isomerase	TPI1	E
Tropomyosin 1 alpha	TPM1	S
Troponin C		S
Troponin I	TNNI3	S
Troponin T2, cardiac	TNNT2	S
Tuberous sclerosis 1	TSC1	G
Tuberous sclerosis 2	TSC2	G
Tumour necrosis factor (TNF) receptor associated factor 1	TRAF1	I
Tumour necrosis factor (TNF) receptor associated factor 2	TRAF2	I
Tumour necrosis factor (TNF) receptor associated factor 3	TRAF3	I
Tumour necrosis factor (TNF) receptor associated factor 4	TRAF4	I
Tumour necrosis factor (TNF) receptor associated factor 5	TRAF5	I
Tumour necrosis factor (TNF) receptor associated factor 6	TRAF6	I
Tumour necrosis factor alpha	TNFA	I
Tumour necrosis factor alpha receptor	TNFAR	I
Tumour necrosis factor beta	TNFB	I
Tumour necrosis factor beta receptor	TNFBR	I
Tumour protein p53	TP53, P53	G
Tumour protein p63	TP63	G
Tyrosine hydroxylase	TH	E
Ubiquitin		G
Ubiquitin B	UBB	G
Ubiquitin C	UBC	G
UDP-glucose pyrophosphorylase		E
UDP-glucuronosyltransferase 1	ugt1d, UGT1	E
UDP-glucuronosyltransferase 2	UGT2	E
Uncoupling protein 1		T
Uncoupling protein 3	UCP3	T
Undulin 1	COL14A1	S
Uridinediphosphate(UDP)-galactose-4-epimerase	GALE	E
Uroporphyrinogen III synthase	UROS	E
Vacuolar proton pump, subunit 1	VPP1	N
Vacuolar proton pump, subunit 3	VPP3	N
Vascular endothelial growth factor	VEGF	G

Vasoactive intestinal polypeptide	VIP	N
Vasoactive intestinal polypeptide receptor	VIPR	N
Vasoinhibitory peptide		G
Vimentin	VIM	I
Vinculin		S
Vitamin D receptor	VDR	G
Von Hippel-Lindau gene	VHL	G
Von Willebrand factor	VWF	T
Werner syndrome helicase	WRN	G
Wiskott-Aldrich syndrome protein	WASP, THC	I
Wolf-Hirschhorn syndrome candidate 1 gene	WHSC1	G
Wolfram syndrome 1 gene	WFS1	S
Xanthine dehydrogenase	XDH	E
Zinc finger protein 3	ZIC3	S

246. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 245.

247. A set according to claim 245 or 246 in which a minority of said probes for listed genes are absent.

248. A set according to claim 245 or 246 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.

249. A set according to claim 245 or 246 in which a limited number of probes are replaced by probes for non-listed genes.

250. A set of probes for a core group of genes according to any of claims 245 to 249 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.

251. A set according to any of claims 245 to 250 consisting of probes for members of a sub-group of the core group.

252. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.

253. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.

254. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.

255. A set according to claim 252 or 253 in which said substrate is a semiconductor microchip.

- 256. A set according to any preceding claim for use in a biological assay for detection of said gene variants.
- 257. A set according to any preceding claim for use in the measurement of differential gene expression levels.
- 258. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
- 259. A medical device including a set according to any of claims 245 to 257 for use in an array for detection of differential gene expression levels.
- 260. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 245) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 245 and 247 to 257 and relating the probe hybridisation pattern to said variations.
- 261. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 246) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 246 to 257 and relating the probe interaction pattern to said variations.
- 262. Use of a set or device according to any of claims 245 to 257 for the prognosis and management of patients suffering from or at risk of experiencing the symptoms or consequences of cardiovascular disease, dysfunction and/or damage.
- 263. Use of a set or device according to any of claims 245 to 257 for predicting likely therapeutic response and adverse events following therapeutic intervention.
- 264. Use of a set or device according to any of claims 245 to 257 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
- 265. Use of a set or device according to any of claims 245 to 257 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
- 266. Use of a set or device according to any of claims 245 to 257 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
- 267. Use of a set or device according to any of claims 245 to 257 for the development of new strategies of therapeutic intervention and in clinical trials.
- 268. Use of a set or device according to any of claims 245 to 257 for construction of and generation of algorithms for patient and healthcare management.
- 269. Use of a set or device according to any of claims 245 to 257 for modelling or assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations
- 270. Use of a set or device according to any of claims 245 to 257 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
- 271. Use of a set or device according to any of claims 245 to 257 for predicting optimum configuration/management of thereapeutic intervention.



272. A method according to claim 260 or 261 in which the identification of gene variants is indicative of a higher risk of developing the symptoms or consequences of cardiovascular disease, dysfunction and/or damage for the patient or individual.
273. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop the symptoms or consequences of cardiovascular disease, dysfunction and/or damage which method comprises:
- i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from the symptoms or consequences of cardiovascular disease, dysfunction and/or damage;
  - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the symptoms or consequences of cardiovascular disease, dysfunction and/or damage;
  - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 245 to 251;
  - iv) calculating the frequencies of these alleles in the samples from i) and ii);
  - v) comparing the frequencies of these alleles in i) and ii);
  - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing the symptoms or consequences of cardiovascular disease, dysfunction and/or damage.
274. A method for assessing whether a given subject will be at risk of developing symptoms, which comprises comparing said subject's genotype with a model generated by the method of claim 273.
275. A method according to any of claims 260, 261, 273 and 274 wherein at least one step is computer-controlled.
276. An assay suitable for use in a method according to any of claims 260, 261, 273 and 274; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 245 to 251 in a biological sample.
277. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing the symptoms or consequences of cardiovascular disease, dysfunction and/or damage; said kit comprising:
- i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 245 or 247 to 251 in a sample of human DNA;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing the symptoms or consequences of cardiovascular disease, dysfunction and/or damage.
278. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing the symptoms or consequences of cardiovascular disease, dysfunction and/or damage; said kit comprising:
- i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core group of genes as defined in any of claims 246 to 251 in an expressed-protein-containing human sample;

- ii) reagents for use in the detection process
- iii) readout indicating the probability of a patient or individual developing the symptoms or consequences of cardiovascular disease, dysfunction and/or damage.

279. A set of probes according to claim 245, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.

280. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to dysfunction, damage or disease of the gastrointestinal tract; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

#### KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME  
T TRANSPORT & STORAGE  
S STRUCTURAL  
I IMMUNITY  
N NERVOUS TRANSMISSION  
G GROWTH & DIFFERENTIATION

#### GASTROINTESTINAL GENE LIST

	HUGO gene symbol	Protein function
11beta hydroxysteroid dehydrogenase 2	HSD11B2	E
17beta hydroxysteroid dehydrogenase 1	HSD17B1	E
17beta hydroxysteroid dehydrogenase 3	HSD17B3	E
17beta hydroxysteroid dehydrogenase 4	HSD17B4	E
17beta hydroxysteroid oxidoreductase		E
2,3-bisphosphoglycerate mutase	BPGM	E
3 beta hydroxysteroid dehydrogenase 2	HSD3B2	E
6-phosphofructo-2-kinase	PFKFB1	E
Acetoacetyl 1-CoA-thiolase	ACAT1	E
Acetoacetyl 2-CoA-thiolase	ACAT2	E
Acetyl CoA carboxylase	ACC	E
Acetyl CoA carboxylase alpha	ACACA	E
Acetylcholine receptor, nicotinic, gamma	CHRNA7	N
Acetylcholinesterase	ACHE	E
Acid phosphatase 2, lysosomal	ACP2	E
Actin, alpha, cardiac	ACTC	S
Actin, alpha, skeletal	ACTA1	S
Actin, alpha, smooth, aortic	ACTA2	S
Actin, beta	ACTB	S
Actin, gamma 2	ACTG2	S
Acyl CoA dehydrogenase, long chain	ACADL	E

Acyl CoA dehydrogenase, medium chain	ACADM	E
Acyl CoA dehydrogenase, short chain	ACADS	E
Acyl CoA dehydrogenase, very long chain	ACADVL	E
Acyl CoA synthetase, long chain, 1	LACS1	E
Acyl CoA synthetase, long chain, 2	LACS2	E
Acyl CoA synthetase, long chain, 4	ACS4	E
Acyl malonyl condensing enzyme		E
Acyl-CoA thioesterase		E
Adaptin, beta 3A	ADTB3A	T
Adenine phosphoribosyltransferase	APRT	T
Adenomatous polyposis coli tumour suppressor gene	APC	G
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N
Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
Adrenocorticotrophic hormone (ACTH) receptor	ACTHR	G
Alanine aminotransferase		T
Alanine-glyoxylate aminotransferase	AGXT	E
Albumin, ALB	ALB	T
Alcohol dehydrogenase 1	ADH1	E
Alcohol dehydrogenase 2	ADH2	E
Alcohol dehydrogenase 3	ADH3	E
Alcohol dehydrogenase 4	ADH4	E
Alcohol dehydrogenase 5	ADH5	E
Alcohol dehydrogenase 6	ADH6	E
Alcohol dehydrogenase 7	ADH7	E
Aldehyde dehydrogenase 1	ALDH1	E
Aldehyde dehydrogenase 2	ALDH2	E
Aldehyde dehydrogenase 5	ALDH5	E
Aldehyde dehydrogenase 6	ALDH6	E
Aldehyde dehydrogenase 7	ALDH7	E
Aldolase A	ALDOA	E

Aldolase B	ALDOB	E
Aldolase C	ALDOC	E
Aldose reductase		T
Aldosterone receptor	MLR	G
Alkaline phosphatase, liver/bone/kidney	ALPL	T
Alpha 2 macroglobulin	A2M	I
alpha1-antitrypsin	PI	E
alpha2-antiplasmin	PLI	E
alpha-actinin 2	ACTN2	G
alpha-actinin 3	ACTN3	G
alpha-amylase		E
alpha-dextrinase		E
alpha-Galactosidase A	GLA	E
alpha-ketoglutarate dehydrogenase		E
alpha-L-Iduronidase	IDUA	E
Aminomethyltransferase	AMT	E
Aminopeptidase P	XPNPEP2	E
Amphiregulin	AREG	G
Amylo-1,6-glucosidase	AGL	E
Angiopoietin 1	ANGPT1	G
Angiopoietin 2	ANGPT2	G
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensin receptor 1	AGTR1	T
Angiotensin receptor 2	AGTR2	T
Angiotensinogen	AGT	E
Antidiuretic hormone receptor	ADHR	T
Antithrombin III	AT3	E
AP-2, alpha	TFAP2A	G
AP-2, beta	TFAP2B	G
AP-2, gamma	TFAP2C	G
Apolipoprotein A I	APOA1	T
Apolipoprotein A II	APOA2	T
Apolipoprotein B	APOB	T
Apolipoprotein C1	APOC1	T
Apolipoprotein C2	APOC2	T
Apolipoprotein C3	APOC3	T
Apolipoprotein D	APOD	T
Apolipoprotein E	APOE	T
Apolipoprotein H	APOH	T
Aquaporin 1	AQP1	T
Aquaporin 2	AQP2	T
Arginine vasopressin	AVP	N
Arginine vasopressin receptor 1A	AVPR1A	N
Arginine vasopressin receptor 1B	AVPR1B	N
Arginine vasopressin receptor 2	AVPR2	N
Arginosuccinate lyase	ASL	E
Arginosuccinate synthetase	ASS	E
Aryl hydrocarbon receptor nuclear translocator	ARNT	T

Arylsulfatase A	ARSA	E
Arylsulfatase B	ARSB	E
Aspartate transaminase		T
Aspartylglucosaminidase	AGA	E
Ataxia telangiectasia gene, AT	ATM	G
ATP/ADP translocase		E
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G
Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
Autoimmune regulator, AIRE	AIRE	I
Azoospermia factor 1	AZF1	G
beta 2 microglobulin	B2M	I
beta-galactosidase	GLB1	E
beta-glucosidase, neutral		E
beta-Glucuronidase	GUSB	E
beta-ketoacyl reductase		E
Bile acid coenzyme A: amino acid N-acyltransferase	BAAT	E
Bile salt export pump	BSEP, PFIC2	T
Bile salt-stimulated lipase	CEL	E
Bilirubin UDP-glucuronosyltransferase		E
Biliverdin reductase		T
Bradykinin receptor B1		I
Bradykinin receptor B2		I
Branched chain keto acid dehydrogenase E1, alpha polypeptide	BCKDHA	E
Branched chain keto acid dehydrogenase E1, beta polypeptide	BCKDHB	E
Brush border guanylyl cyclase		E
Ca(2+) transporting ATPase, fast twitch	ATP2A1	T
Ca(2+) transporting ATPase, slow twitch	ATP2A2	T
Cadherin E	CDH1	G
Cadherin EP		G
Cadherin N	CDH2	G
Cadherin P	CDH3	G
Calcitonin/Calcitonin gene-related peptide alpha	CALCA	N
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha-1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha-1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha-1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha-1E (CACNL1A6)	CACNA1E	N

Calcium channel, voltage-dependent, Alpha-2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N
Calcium channel, voltage-dependent, T-type		N
Calcium sensing receptor	CASR	T
Calmodulin 1	CALM1	G
Calmodulin 2	CALM2	G
Calmodulin 3	CALM3	G
Calmodulin dependant kinase		T
Calmodulin-dependant protein kinase II	CAMK2A	G
Calnexin	CANX	G
Canalicular multispecific organic anion transporter	CMOAT	T
Carbamoylphosphate synthetase 1	CPS1	E
Carbamoylphosphate synthetase 2	CPS2	E
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Carboxylesterase 1	CES1	E
Carboxypeptidase	CPN	E
Carnitine acylcarnitine translocase	CACT	E
Carnitine palmitoyltransferase I	CPT1A	E
Carnitine palmitoyltransferase II	CPT2	E
Carnitine transporter protein	CDSP, SCD	T
Cartilage-hair hypoplasia gene	CHH	N
Catalase	CAT	I
Cathepsin B		E
Cathepsin D		E
Cathepsin E		E
Cathepsin G	CTSG	E
Cathepsin H		E
Cathepsin K	CTSK	E
Cathepsin L		E
Cathepsin S		E
CD1	CD1	I
CD4	CD4	I
Cell adhesion molecule, intercellular, ICAM	ICAM1	G
Cell adhesion molecule, leukocyte-endothelial, LECAM (CD62)	LECAM1	G
Cell adhesion molecule, liver, LCAM	LCAM	G
Cell adhesion molecule, neural, NCAM1	NCAM1	G
Cell adhesion molecule, neural, NCAM120	NCAM120	G
Cell adhesion molecule, neural, NCAM2	NCAM2	G
Cell adhesion molecule, platelet-endothelial,	PECAM1	G

PECAM		
Cell adhesion molecule, vascular, VCAM	VCAM1	G
c-erbB2	ERBB2	G
c-erbB3	ERBB3	G
c-erbB4	ERBB4	G
Ceruloplasmin precursor	CP	E
Chemokine receptor CCR2	CCR2	I
Chemokine receptor CCR3	CCR3	I
Chemokine receptor CCR5	CCR5	I
Chemokine receptor CXCR4	CXCR4	I
Chitotriosidase	chit	E
Chloride channel 5	CLCN5	S
Chloride channel KB	CLCNKB	S
Cholecystokinin	CCK	N
Cholecystokinin B receptor	CCKBR	N
Cholestasis, progressive familial intrahepatic 1 gene	FIC1	G
Cholesterol ester hydroxylase		E
Choline acetyltransferase	CHAT	E
Chromogranin A	CHGA	G
Chymotrypsinogen		E
Citrate synthase		E
Clathrin		T
Clusterin	CLU	G
CoA transferase		E
Cockayne syndrome gene, CKN1	CKN1	G
Collagen I alpha 1	COL1A1	S
Collagen I alpha 2	COL1A2	S
Collagen II alpha 1	COL2A1	S
Collagen III alpha 1	COL3A1	S
Collagen IV alpha 1	COL4A1	S
Collagen IV alpha 2	COL4A2	S
Collagen IV alpha 3	COL4A3	S
Collagen IV alpha 4	COL4A4	S
Collagen IV alpha 5	COL4A5	S
Collagen IV alpha 6	COL4A6	S
Collagen IX alpha 2	COL9A2, EDM2	S
Collagen IX alpha 3	COL9A3	S
Collagen receptor	COLR	S
Collagen V alpha 1	COL5A1	S
Collagen V alpha 2	COL5A2	S
Collagen VI alpha 1	COL6A1	S
Collagen VI alpha 2	COL6A2	S
Collagen VI alpha 3	COL6A3	S
Collagen VII alpha 1	COL7A1	S
Collagen X alpha 1	COL10A1	S
Collagen X alpha 1	COL11A1	S
Collagen XI alpha 2	COL11A2	S

Collagen XVII alpha 1	COL17A1	S
Colony-stimulating factor 1	CSF1	G
Complement component C1 inhibitor	C1NH	I
Complex I		E
Complex II		E
Complex III		E
Corticotrophin-releasing hormone	CRH	T
Corticotrophin-releasing hormone receptor	CRHR1	T
C-reactive protein CRP		I
Creb binding protein	CREBBP	G
Cu <sup>2+</sup> transporting ATPase beta polypeptide	ATP7B	E
Cubilin	CUBN	T
Cyclic AMP-dependent protein kinase	PKA	E
Cyclic nucleotide phosphodiesterase 1B	PDE1B	E
Cyclic nucleotide phosphodiesterase 1B1	PDE1B1	E
Cyclic nucleotide phosphodiesterase 2A3	PDE2A3	E
Cyclic nucleotide phosphodiesterase 3A	PDE3A	E
Cyclic nucleotide phosphodiesterase 3B	PDE3B	E
Cyclic nucleotide phosphodiesterase 4A	PDE4A	E
Cyclic nucleotide phosphodiesterase 4C	PDE4C	E
Cyclic nucleotide phosphodiesterase 5A	PDE5A	E
Cyclic nucleotide phosphodiesterase 6A	PDE6A	E
Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E
Cyclic nucleotide phosphodiesterase 8	PDE8	E
Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclin F	CCNF	G
Cyclin-dependent kinase 2	CDK2	G
Cyclin-dependent kinase inhibitor 1C (P57, KIP2)	CDKN1C	G
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
CYP11A1	CYP11A1	E
CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E



Dopamine receptors D2	DRD2	N
Dopamine receptors D3	DRD3	N
Dopamine receptors D4	DRD4	N
Dopamine receptors D5	DRD5	N
Dynamin	DNM1	G
Dynein		G
Dystrophia myotonica	DM, DMPK	E
Dystrophia myotonica, atypical	DM2	E
Dystrophin	DMD	S
EB1		G
Elastase 1	ELAS1	E
Elastase 2	ELAS2	E
Electron-transferring-flavoprotein alpha	ETFA	T
Electron-transferring-flavoprotein beta	ETFB	T
Electron-transferring flavoprotein dehydrogenase	ETFDH	E
Endothelin 1	EDN1	N
Endothelin 2	EDN2	N
Endothelin 3	EDN3	N
Endothelin converting enzyme	ECE1	N
Endothelin receptor type A	EDNRA	N
Endothelin receptor type B	EDNRB	N
Enolase	ENO1	E
Enoyl CoA isomerase		E
Enoyl CoA reductase		E
Enteric lipase		T
Enterokinase	PRSS7, ENTK	E
Ephrin receptor tyrosine kinase A	EPHA	G
Ephrin receptor tyrosine kinase B	EPHB	G
Epidermal growth factor	EGF	G
Epidermal growth factor receptor	EGFR	G
Erythrocyte membrane protein band 4.1	EPB41	S
Erythropoietin	EPO	I
Excision repair complementation group 2 protein	ERCC2	E
Excision repair complementation group 2 protein	ERCC3	E
Eyes absent 1	EYA1	G
Faciogenital dysplasia	FGD1, FGDY	T
Factor 1 (No. one)	F1	I
Factor B, properdin		I
Factor D		I
Factor H	HF1	I
Factor I (letter I)	IF	I
Factor III	F3	I
Factor IX	F9	I
Factor V	F5	I
Factor VII	F7	I

Factor VIII	F8	I
Factor X	F10	I
Factor XI	F11	I
Factor XII	F12	I
Factor XIII A & B	F13A & F13B	I
FADH dehydrogenase		E
Fanconi anemia, complementation group A	FANCA	T
Fanconi anemia, complementation group C	FANCC	T
Fanconi anemia, complementation group D	FANCD	T
Fatty acid binding proteins FABP1		T
Fatty acid binding proteins FABP2	FABP2	T
Fatty acid binding proteins FABP3		T
Fatty acid binding proteins FABP4		T
Fatty acid binding proteins FABP5		T
Fatty acid binding proteins FABP6		T
Ferritin, H subunit		T
Ferritin, L subunit	FTL	T
Fibroblast growth factor	FGF1	G
Fibroblast growth factor receptor 1	FGFR1	G
Fibroblast growth factor receptor 2	FGFR2	G
Fibroblast growth factor receptor 3	FGFR3	G
Fibronectin precursor	FN1	G
Flavin-containing monooxygenase 1	FMO1	E
Flavin-containing monooxygenase 2	FMO2	E
Flavin-containing monooxygenase 3	FMO3	E
Flavin-containing monooxygenase 4	FMO4	E
Folic acid receptor	FOLR	G
Follicle stimulating hormone receptor	FSHR, ODG1	G
Follicle stimulating hormone, FSH	FSHB	G
Forkhead transcription factor 10	FKHL10	G
Forkhead transcription factor 14	FKHL14	G
Fragile site, folic acid type, rare, fra(X) A	FRAXA	N
Fructose-1,6-diphosphatase	FBP1	E
Fucosidase alpha-L-1	FUCA1	E
Fucosidase alpha-L-2		E
Fucosyltransferase 2	FUT2	T
Fucosyltransferase 3	FUT3	T
Fumarase	FH	E
G/T mismatch binding protein	GTBP, MSH6	G
Galactocerebrosidase	GALC	E
Galactose 1-phosphate uridyl-transferase	GALT	E
Galactosyltransferase 1	GT1	G
Galactosyltransferase, alpha 1,3	GGTA1	G
Galactosyltransferase, beta 3	B3GALT	G
Galanin	GAL	N
Galanin receptor	GALNR1	N
Gamma-glutamyltransferase 1	GGT1	T
Gamma-glutamyltransferase 2	GGT2	T

Gap junction protein beta 1	GJB1	T
Gastric inhibitory polypeptide GIP	GIP	T
Gastric inhibitory polypeptide receptor, GIPR	GIPR	T
Gastric Intrinsic factor, GIF	GIF	E
Gastric lipase, LIPF		T
Gastrin	GAS	G
Gastrin releasing peptide	GRP	T
Gastrin releasing peptide receptor	GRPR	T
Glial-cell derived neurotrophic factor (GDNF) receptor		N
Glial-cell derived neurotrophic factor, GDNF	GDNF	N
Glucagon receptor	GCGR	G
Glucagon synthase		T
Glucagon-like peptide receptor 1	GLP1R	G
Glucokinase	GCK	E
Glucose-6-phosphatase	G6PC	E
Glucose-6-phosphatase translocase	G6PT1	E
Glucose-6-phosphate dehydrogenase	G6PD	E
Glucosidase, acid alpha	GAA	E
Glutamate dehydrogenase	GLUD1	E
Glutamine synthase		E
Glutamine transporter		T
Glutathione	GSH	T
Glutathione peroxidase, GPX2	GPX2	E
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
Glycerol kinase	GK	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
Glycine dehydrogenase	GLDC	E
Glycogen branching enzyme	GBE1	E
Glycogen phosphorylase	PYGL	E
Glycogen synthase 1 (muscle)	GLYS1	E
Glycogen synthase 2 (liver)	GYS2	E
Glycosyltransferases, ABO blood group	ABO	E
Gonadotropin releasing hormone	GNRH	G
Goosecoid GSC		G
Growth arrest-specific homeobox	GAX	G
Growth hormone receptor	GHR	G
Guanylin	GUCA2	T
H(+), K(+) - ATPase	ATP4B	N
Haem oxygenase		T
Haemoglobin alpha 1	HBA1	T
Haemoglobin alpha 2	HBA2	T
Haemoglobin beta	HBB	T
Haemoglobin delta	HBD	T
Haemoglobin gamma A	HBG1	T

Haemoglobin gamma B	HBG2	T
Haemoglobin gamma G	HBGG	T
Heat shock protein, HSP60		I
Heat shock protein, HSP70		I
Heat shock protein, HSP90		I
Heat shock protein, HSPA1		I
Heat shock protein, HSPA2		I
Heparan sulfamidase		E
Heparin binding epidermal growth factor	HBEGF	G
Heparin Cofactor II	HCF2	I
Hepatic nuclear factor-3-beta	HNF3B	E
Hepatic nuclear factor-4-alpha	HNF4A	E
Hepatitis B virus integration site 1	HVBS1	I
Hepatitis B virus integration site 2	HVBS6	I
Hepatocyte growth factor	HGF	G
Hermansky-pudlak syndrome gene	HPS	T
Hexokinase 1	HK1	E
Hexokinase 2	HK2	E
Hexosaminidase A	HEXA,TSD	E
Hexosaminidase B	HEXB	E
Histamine receptors, H1		N
Histamine receptors, H2		N
Histamine receptors, H3		N
Histatin 1		I
Histatin 2		I
Histatin 3	HTN3	I
HLA-B associated transcript 1	BAT1	I
HMG-CoA lyase	HMGCL	E
HMG-CoA reductase	HMGCR	E
HMG-CoA synthase	HMGCS2	E
Holocarboxylase synthetase	HLCS	E
Hormone-sensitive lipase	HSL	E
Hydroxyacyl glutathione hydrolase	HAGH	E
Hypoxanthine-guanine phosphoribosyltransferase, HGPRT	HPRT	E
IC7 A and B		I
Iduronate 2 sulphotase	IDS	E
Immunoglobulin E (IgE) responsiveness gene	IGER	I
Immunoglobulin E (IgE) serum concentration regulator gene	IGES	I
Immunoglobulin gamma (IgG) 2	IGHG2	I
Immunoglobulin heavy mu chain	IGHM	I
Immunoglobulin J polypeptide	IGJ	I
Immunoglobulin kappa constant region	IGKC	I
Immunoglobulin kappa variable region	IGKV	I
Inhibin, alpha	INHA	G
Inhibin, beta A	INHBA	G
Inhibin, beta B	INHBB	G

Inhibin, beta C	INHBC	G
Inositol 1,4,5-triphosphate receptor 3	ITPR3	G
Insulin	INS	G
Insulin receptor	INSR	G
Insulin-like growth factor 1	IGF1	G
Insulin-like growth factor 1 receptor	IGF1R	G
Insulin-like growth factor 2	IGF2	G
Insulin-like growth factor 2 receptor	IGF2R	G
Integrin beta 1	ITGB1	G
Integrin beta 2	ITGB2	G
Integrin beta 3	ITGB3	G
Integrin beta 6	ITGB6	G
Integrin, alpha M	ITGAM	G
Integrin, alpha X	ITGAX	G
Inter-alpha-trypsin inhibitor, IAT1		E
Interferon alpha	IFNA1	I
Interferon beta	IFNB	I
Interferon gamma	IFNG	I
Interferon gamma receptor 1	IFNGR1	I
Interferon gamma receptor 2	IFNGR2	I
Interferon regulatory factor 1	IRF1	I
Interferon regulatory factor 4	IRF4	I
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) 10	IL10	I
Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I
Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I

Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
Intestinal alkaline phosphatase IAP		T
Islet amyloid polypeptide	IAPP	N
Isocitrate dehydrogenase		E
Isovaleric acid CoA dehydrogenase	IVD	E
Kallikrein 3	KAK3	I
Kallman syndrome gene 1	KAL1	G
Ketohexokinase	KHK	E
ketolase		E
Kininogen, High molecular weight	KNG	I
Kynurenine hydroxylase		E
Kynureninase		E
Lactase		E
Laminin 5, alpha 3	LAMA3	G
Laminin 5, beta 3	LAMB3	G
Laminin 5, gamma 2	LAMC2	G
Laminin M	LAMM	G
Laminin receptor 1	LAMR1	G
Latent transforming growth factor-beta binding protein 2	LTBP2	G
Lecithin-cholesterol acyltransferase	LCAT	E
Leptin	LEP	G
Leptin receptor	LEPR	G
Leukocyte-specific transcript 1	LST-1	I
Leukotriene A4 hydrolase		I
Leukotriene A4 synthase	LTA4S	E
Leukotriene B4 receptor		I
Leukotriene B4 synthase	LTB4S	E
Leukotriene C4 receptor		I
Leukotriene C4 synthase	LTC4S	E
Leukotriene D4/E4 receptor		I
LIM homeobox protein 1	LHX1	G
LIM homeobox transcription factor 1, beta	LMX1B	G
Lipoamide dehydrogenase	OGDH	E
Lipoprotein lipase	LPL	I
Lipoprotein receptor, Low Density	LDLR	T
Lipoprotein, High Density	HDLDT1	T
Lipoprotein, Intermediate Density		T
Lipoprotein, Low Density 1		T
Lipoprotein, Low Density 2		T
Lipoprotein, Very Low Density	VLDLR	T
Low density lipoprotein receptor-related protein precursor	LRP	T
Lymphoid enhancer-binding factor	LEF-1	G
Lysosomal acid lipase	LIPA	E
Lysozyme	LYZ	I

MAD (mothers against decapentaplegic, Drosophila) homologue 4	MADH4	G
MADS box transcription-enhancer factor 2A	MEF2A	G
MADS box transcription-enhancer factor 2B	MEF2B	G
MADS box transcription-enhancer factor 2C	MEF2C	G
MADS box transcription-enhancer factor 2D	MEF2D	G
Malonyl CoA decarboxylase		E
Malonyl CoA transferase		E
Maltase-glucoamylase		E
Mannosidase, alpha B lysosomal	MANB	E
Marenostrin	MEFV	T
MAX-interacting protein 1	MXI1	G
MEK kinase, MEKK		E
Melanocortin 2 receptor	MC2R	T
Melanocortin 4 receptor	MC4R	T
Menin	MEN1	G
Metallothionein		T
Mevalonate kinase	MVK	E
MHC Class I: A		I
MHC Class I: B		I
MHC Class I: C		I
MHC Class I: LMP-2, LMP-7		I
MHC Class I: Tap1	ABCR, TAP1	I
MHC Class II: DP	HLA-DPB1	I
MHC Class II: DQ		I
MHC Class II: DR		I
MHC Class II: Tap2	TAP2, PSF2	I
MHC Class II:Complementation group A	MHC2TA	I
MHC Class II:Complementation group B	rfxank	I
MHC Class II:Complementation group C	RFX5	I
MHC Class II:Complementation group D	RFXAP	I
Microsomal triglyceride transfer protein	MTP	T
Mitochondrial trifunctional protein, alpha subunit	HADHA	E
Mitochondrial trifunctional protein, beta subunit	HADHB	E
Molybdenum cofactor synthesis 1	MOCS1	E
Molybdenum cofactor synthesis 2	MOCS2	E
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Motilin	MLN	G
Msh homeobox homolog 2	MSX2	G
Mucin 18	MUC18	T
Mucin, MUC2		T
Mucin, MUC5AC		T
Mucin, MUC6		T
Mucopolidoses	GNPTA	E
Mulibrey nanism	MUL	T
Muscarinic receptor, M1	CHRM1	N

Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Muscle phosphorylase	PYGM	E
Mutated in colorectal cancers, MCC	MCC	G
MutL homolog 1	MLH1	G
MutS homolog 2	MSH2	G
MutS homolog 3	MSH3	G
Myoglobin		T
Myosin 15	MYO15	S
Myosin 5A	MYO5A	S
Myosin 6	MYO6	S
Myosin 7A	MYO7A	S
Myosin, cardiac	MYH7	S
Myosin, light chain 2	MYL2	S
Myosin, light chain 3	MYL3	S
Myotubularin	MTM1	S
Na <sup>+</sup> , K <sup>+</sup> ATPase, alpha	ATP1A1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 1	ATP1B1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 2	ATP1B2	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 3	ATP1B3	G
Na <sup>+</sup> /H <sup>+</sup> exchanger 1	NHE1	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 2	NHE2	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 3	NHE3	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 4	NHE4	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 5	NHE5	T
Na <sup>+</sup> -coupled glucose/galactose transporter		T
N-acetylgalactosamine-6-sulfate sulfatase	GALNS	E
N-acetylglucosamine-6-sulfatase	GNS	E
N-acetylglucosaminidase, alpha	NAGLU	E
NADH dehydrogenase		E
NADH dehydrogenase (ubiquinone) Fe-S protein 1	NDUFS1	E
NADH dehydrogenase (ubiquinone) Fe-S protein 4	NDUFS4	E
NADH dehydrogenase (ubiquinone) flavoprotein 1	NDUFV1	E
NADH-cytochrome b5 reductase	DIA1	E
NADPH-dependent cytochrome P450 reductase	POR	E
NB6		I
Nephrolithiasis 2	NPHL2	T
Nephronophthisis 1	NPHP1	T
Nephronophthisis 2	NPHP2	T
Nephrosis 1	NPHS1	T
Nerve growth factor	NGF	G
Nerve growth factor receptor	NGFR	G



Neuraminidase sialidase	NEU	T
Neurofibromin 1	NF1	G
Neurofibromin 2	NF2	G
Neurokinin A	NKNA	N
Neurokinin B	NKNB	N
Neurotensin	NTS	N
Neurotensin receptor	NTSR1	N
Notch ligand - jagged 1	JAG1, AGS	G
Nuclear factor I-kappa-B-like gene	IKBL	I
Oncogene ERB		G
Oncogene ERB2		G
Oncogene ERBA		G
Oncogene ERBAL2		G
Oncogene GLI1	GLI	G
Oncogene GLI2	GLI2	G
Oncogene GLI3	GLI3	G
Oncogene met	MET	G
Oncogene myb	MYB	G
Oncogene myc	MYC	G
Oncogene n-myc		G
Oncogene ret	RET	G
Oncogene r-myc		G
Oncogene sis	PDGFB	G
Oncogene spi1		G
Oncogene src		G
Oncogene v-Ki-ras2	KRAS2	G
Orexin	OX	G
Orexin 1 receptor	OX1R	G
Orexin 2 receptor	OX2R	G
Ornithine transcarbamoylase	OTC, NME1	E
Osteopontin	OPN	G
Paired box homeotic gene 2	PAX2	G
Paired box homeotic gene 3	PAX3	G
Paired box homeotic gene 6	PAX6	G
Paired box homeotic gene 8	PAX8	G
Palmitoyl-protein thioesterase	PPT	T
Pancreatic amylase		E
Pancreatic colipase		T
Pancreatic lipase	PNLIP	E
Pancreatic lipase related protein 1	PLRP1	E
Pancreatic lipase related protein 2	PLRP2	E
Paraoxonase PON1	PON1	E
Paraoxonase PON2	PON2	E
Paraoxonase PON3		E
Parathyroid hormone	PTH	G
Parathyroid hormone receptor	PTHrP	G
Parathyroid hormone related-peptide	PTHrP	G
Parathyroid hormone-like hormone	PTHrP	G

Parvalbumin	PVALB	G
Patched (Drosophila) homolog, PTCH	PTCH	G
Pepsin		T
Pepsinogen		E
Peptidases A		T
Peptidases B		T
Peptidases C		T
Peptidases D	PEPD	T
Peptidases E		T
Peptidases S		T
Peroxidase, salivary	SAPX	E
Peroxisomal membrane protein 1	PXMP1	S
Peroxisomal membrane protein 3	PXMP3	T
Peroxisome biogenesis factor 1	PEX1	T
Peroxisome biogenesis factor 19	PEX19	T
Peroxisome biogenesis factor 6	PEX6	T
Peroxisome biogenesis factor 7	PEX7	T
Peroxisome receptor 1	PXR1	T
Phenylalanine monooxygenase		E
Phosphatase & tensin homolog	PTEN	G
Phosphate regulating gene with homologies to endopeptidases on the X chromosome	PHEX	G
Phosphoenolpyruvate carboxykinase	PCK1	E
Phosphofructokinase, liver	PFKL	E
Phosphofructokinase, muscle	PFKM	E
Phosphoglucomutase		E
Phosphoglucose isomerase	GPI	E
Phosphoglycerate kinase 1	PGK1	E
Phosphoglycerate mutase 2	PGAM2	E
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C alpha		I
Phospholipase C beta		I
Phospholipase C delta	PLCD1	I
Phospholipase C epsilon		I
Phospholipase C gamma	PLCG1	I
Phosphomannomutase 2	PMM2	G
Phosphomannomutase-2	PMM2	T
Phosphomannose isomerase-1, PMI1	MPI	T
Phosphoribosyl pyrophosphate synthetase	PRPS1	E
Phosphorylase kinase deficiency, liver	PHK	E
Phosphorylase kinase, alpha 1 (muscle)	PHKA1	E

Phosphorylase kinase, alpha 2	PHKA2	E
Phosphorylase kinase, beta	PHKB	E
Phosphorylase kinase, delta		E
Phosphorylase kinase, gamma 2	PHKG2	E
Plasminogen	PLG	E
Plasminogen activator inhibitor 1	PAI1	E
Plasminogen activator inhibitor 2	PAI2	E
Plasminogen activator receptor, Urokinase	UPAR; PLAUR	S
Plasminogen activator, Tissue	PLAT; TPA	E
Plasminogen activator, Urokinase	UPA; PLAU	E
Platelet derived growth factor	PDGF	G
Platelet derived growth factor receptor	PDGFR	G
Platelet monamine oxidase		T
Platelet-activating factor receptor	PAFR	I
Polycystic kidney and hepatic disease 1	PKHD1	T
Polycystin 1	PKD1	T
Polycystin 2	PKD2	T
Polymorphonuclear elastase		T
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium inwardly-rectifying channel J11	KCNJ11	N
Potassium voltage-gated channel E1	KCNE1	N
Prekallikrein		I
Preproenkephalin	PENK	N
Preproglucagon	GCG;GLP1; GLP2	G
Preproglucagon		T
Preproinsulin		T
Procollagen N-protease		E
Proline dehydrogenase	PRODH	E
Proline-rich protein BstNI subfamily 1	PRB1	S
Proline-rich protein BstNI subfamily 3	PRB3	S
Proline-rich protein BstNI subfamily 4	PRB4	S
Prolyl-4-hydroxylase		E
Pro-melanin-concentrating hormone	PMCH	G
Proopiomelanocortin	POMC	N
Prosaposin	PSAP	N
Prostacyclin synthase		I
Prostaglandin 15-OH dehydrogenase	HGPD; PGDH	I
Prostaglandin D - DP receptor		I
Prostaglandin E1 receptor		I
Prostaglandin E2 receptor		I
Prostaglandin E3 receptor		I
Prostaglandin F - FP receptor		I
Prostaglandin F2 alpha receptor		I
Prostaglandin I2 receptor		T
Prostaglandin IP receptor		I
Protease inhibitor 1		T
Protective protein for beta-galactosidase	PPGB	E
Protein C	PROC	I

Protein C inhibitor	PCI	I
Protein kinase B	PRKB	
Protein S	PROS1	I
Protein tyrosine phosphatase, non-receptor type 12	PTPN12	G
Prothrombin precursor	F2	I
Pterin-4-alpha-carbinolamine	PCBD	
Pyruvate carboxylase	PC	E
Pyruvate decarboxylase	PDHA	E
Pyruvate kinase	PKLR	E
Quinoid dihydropteridine reductase	QDPR	E
Renal glutaminase		T
Renin	REN	E
Replication factor C	RFC2	E
Retinoblastoma 1	RB1	G
Retinol binding protein 1		T
Retinol binding protein 2		T
Retinoschisis, X-linked, juvenile	RS	G
RIGUI	RIGUI	G
SA homolog	SAH	G
Salivary amylase, AMY1		T
SAP (SLAM-associated protein)	SH2D1A	I
Secretin	SCT	T
Secretin receptor, SCTR	SCTR	T
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Solute carrier family 10 (sodium/bile acid cotransporter family), member 1	SLC10A1	T
Solute carrier family 10 (sodium/bile acid cotransporter family), member 2	SLC10A2	T
Solute carrier family 12, member 1	SLC12A1	T

Solute carrier family 12, member 2	SLC12A2	T
Solute carrier family 12, member 3	SLC12A3	T
Solute carrier family 14, member 2	SLC14A2	T
Solute carrier family 15 (H <sup>+</sup> /peptide transporter, intestinal), member 1	SLC15A1	T
Solute carrier family 15 (H <sup>+</sup> /peptide transporter, kidney), member 2	SLC15A2	T
Solute carrier family 16 (monocarboxylate transporter), member 1	SLC16A1	T
Solute carrier family 16 (monocarboxylate transporter), member 7	SLC16A7	T
Solute carrier family 17, member 1	SLC17A1	T
Solute carrier family 17, member 2	SLC17A2	T
Solute carrier family 2 (facilitated glucose transporter), member 1	SLC2A1	T
Solute carrier family 2 (facilitated glucose transporter), member 2	SLC2A2	T
Solute carrier family 2 (facilitated glucose transporter), member 3	SLC2A3	T
Solute carrier family 2 (facilitated glucose transporter), member 4	SLC2A4	T
Solute carrier family 2 (facilitated glucose transporter), member 5	SLC2A5	T
Solute carrier family 21, member 2	SLC21A2	T
Solute carrier family 21, member 3	SLC21A3	T
Solute carrier family 22, member 1	SLC22A1	T
Solute carrier family 22, member 2	SLC22A2	T
Solute carrier family 22, member 5	SLC22A5	T
Solute carrier family 3 (facilitated glucose transporter), member 1	SLC3A1	T
Solute carrier family 4 (anion exchanger), member 1	SLC4A1	T
Solute carrier family 4 (anion exchanger), member 2	SLC4A2	T
Solute carrier family 4 (anion exchanger), member 3	SLC4A3	T
Solute carrier family 5 (sodium/glucose transporter), member 1	SLC5A1	T
Solute carrier family 5 (sodium/glucose transporter), member 2	SLC5A2	T
Solute carrier family 5 (sodium/glucose transporter), member 5	SLC5A5	T
Solute carrier family 5, member 3	SLC5A3	T
Solute carrier family 6 (GAMMA-AMINOBUTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter	SLC6A2	T

transporter, noradrenaline), member 2		
Solute carrier family 6, member 6	SLC6A6	T
Solute carrier family 7(amino acid transporter), member 1	SLC7A1	T
Solute carrier family 7(amino acid transporter), member 2	SLC7A2	T
Solute carrier family 7(amino acid transporter), member 7	SLC7A7	T
Somatostatin	SST	N
Somatostatin receptor, SSTR1	SSTR1	N
Somatostatin receptor, SSTR2	SSTR2	G
Somatostatin receptor, SSTR3	SSTR3	N
Somatostatin receptor, SSTR4	SSTR4	N
Somatostatin receptor, SSTR5	SSTR5	N
Sphingomyelinase	SMPD1	E
Steroid 5 alpha reductase 1	SRD5A1	E
Steroid 5 alpha reductase 2	SRD5A2	E
Sterol carrier protein 2	SCP2	T
Substance P		N
Succinyl CoA synthase		E
Sucrase		E
Sucrase-isomaltase	SI	T
Superoxide dismutase 1	SOD1	E
Surfeit 1	SURF1	G
Talin	TLN	G
Talin, TLN		S
TATA binding protein	TBP	G
T-BOX 1	TBX1	G
T-BOX 2	TBX2	G
T-BOX 3	TBX3	G
Thiolase, peroxisomal		E
Thrombin receptor	F2R	I
Thrombopoietin	THPO	G
Thromboxane A synthase 1	TBXAS1	I
Tip-associated protein	TAP	I
Topoisomerase I		E
Torticollis, keloids, cryptorchidism and renal dysplasia gene	TKCR	G
Transacylase		E
Transcobalamin 1, TCN1		T
Transcobalamin 2, TCN2	TCN2	T
Transcription factor 1, hepatic	TCF1	G
Transcription factor 2, hepatic	TCF2	G
Transferrin	TF	G
Transferrin receptor	TFRC	G
Transforming growth factor, beta 2	TGFB2	G
Transforming growth factor, beta induced	TGFBI	G
Transforming growth factor, beta receptor 2	TGFBR2	G

Transglutaminase 4	TGM4	G
Transketolase	TKT	E
Transketolase-like 1	TKTL1	E
Translocation in renal carcinoma on chromosome 8 gene	TRC8	G
Transthyretin	TTR	T
Trehalase		T
Triosephosphate isomerase	TPI1	E
Trypsin inhibitor		E
Trypsinogen 1	TRY1	E
Trypsinogen 2	TRY2	E
Trypsinogen activation peptide		T
Tuberous sclerosis 1	TSC1	G
Tuberous sclerosis 2	TSC2	G
Tumour necrosis factor (TNF) receptor associated factor 1	TRAF1	I
Tumour necrosis factor (TNF) receptor associated factor 2	TRAF2	I
Tumour necrosis factor (TNF) receptor associated factor 3	TRAF3	I
Tumour necrosis factor (TNF) receptor associated factor 4	TRAF4	I
Tumour necrosis factor (TNF) receptor associated factor 5	TRAF5	I
Tumour necrosis factor (TNF) receptor associated factor 6	TRAF6	I
Tumour necrosis factor alpha	TNFA	I
Tumour necrosis factor alpha receptor	TNFAR	I
Tumour necrosis factor beta	TNFB	I
Tumour necrosis factor beta receptor	TNFBR	I
Tumour protein p53	TP53, P53	G
Tumour protein p63	TP63	G
Tumour suppressor gene DRA	DRA	I
Tyrosinase	TYR	E
UDP-glucose pyrophosphorylase		E
UDP-glucuronosyltransferase 1	ugt1d, UGT1	E
UDP-glucuronosyltransferase 2	UGT2	E
Uridinediphosphate(UDP)-galactose-4-epimerase	GALE	E
Uroporphyrinogen decarboxylase	UROD	E
Uroporphyrinogen III synthase	UROS	E
Vasoactive intestinal polypeptide	VIP	N
Vasoactive intestinal polypeptide receptor	VIPR	N
Vasoinhibitory peptide		G
Villin		S
Von Hippel-Lindau gene	VHL	G
Von Willebrand factor	VWF	T
Wiskott-Aldrich syndrome protein	WASP, THC	I

Wolf-Hirschhorn syndrome candidate 1 gene	WHSC1	G
Wolfram syndrome 1 gene	WFS1	S
Xanthine dehydrogenase	XDH	E
Xeroderma pigmentosum, complementation group A	XPA	E
Xeroderma pigmentosum, complementation group B	XPB	E
Xeroderma pigmentosum, complementation group C	XPC	E
Xeroderma pigmentosum, complementation group D		E
Xeroderma pigmentosum, complementation group E		E
Xeroderma pigmentosum, complementation group F	XPF	E
Xeroderma pigmentosum, complementation group G	ERCC5	E
Zinc finger protein 3	ZIC3	S

281. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 280.

282. A set according to claim 280 or 281 in which a minority of said probes for listed genes are absent.

283. A set according to claim 280 or 281 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.

284. A set according to claim 280 or 281 in which a limited number of probes are replaced by probes for non-listed genes.

285. A set of probes for a core group of genes according to any of claims 280 to 284 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.

286. A set according to any of claims 280 to 285 consisting of probes for members of a sub-group of the core group.

287. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.

288. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.



289. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.
290. A set according to claim 287 or 288 in which said substrate is a semiconductor microchip.
291. A set according to any preceding claim for use in a biological assay for detection of said gene variants.
292. A set according to any preceding claim for use in the measurement of differential gene expression levels.
293. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
294. A medical device including a set according to any of claims 280 to 292 for use in an array for detection of differential gene expression levels.
295. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 280) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 280 and 282 to 292 and relating the probe hybridisation pattern to said variations.
296. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 281) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 281 to 292 and relating the probe interaction pattern to said variations.
297. Use of a set or device according to any of claims 280 to 292 for the prognosis and management of patients suffering from or at risk of experiencing the symptoms and consequences of dysfunction, damage or disease of the gastrointestinal tract.
298. Use of a set or device according to any of claims 280 to 292 for predicting likely therapeutic response and adverse events following therapeutic intervention.
299. Use of a set or device according to any of claims 280 to 292 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
300. Use of a set or device according to any of claims 280 to 292 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
301. Use of a set or device according to any of claims 280 to 292 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
302. Use of a set or device according to any of claims 280 to 292 for the development of new strategies of therapeutic intervention and in clinical trials.
303. Use of a set or device according to any of claims 280 to 292 for construction of and generation of algorithms for patient and healthcare management.
304. Use of a set or device according to any of claims 280 to 292 for modelling or assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations.

305. Use of a set or device according to any of claims 280 to 292 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
306. Use of a set or device according to any of claims 280 to 292 for predicting optimum configuration/management of thereapeutic intervention.
307. A method according to claim 295 or 296 in which the identification of gene variants is indicative of a higher risk of developing dysfunction, damage or disease of the gastrointestinal tract or of experiencing the symptoms and consequences of dysfunction, damage or disease of the gastrointestinal tract for the patient or individual.
308. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop symptoms and consequences of dysfunction, damage or disease of the gastrointestinal tract which method comprises:
- i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from symptoms and consequences of dysfunction, damage or disease of the gastrointestinal tract;
  - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the symptoms and consequences of dysfunction, damage or disease of the gastrointestinal tract;
  - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 280 to 286;
  - iv) calculating the frequencies of these alleles in the samples from i) and ii);
  - v) comparing the frequencies of these alleles in i) and ii);
  - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing symptoms and consequences of dysfunction, damage or disease of the gastrointestinal tract.
309. A method for assessing whether a given subject will be at risk of developing symptoms, which comprises comparing said subject's genotype with a model generated by the method of claim 308.
310. A method according to any of claims 295, 296, 308 and 309 wherein at least one step is computer-controlled.
311. An assay suitable for use in a method according to any of claims 295, 296, 308 and 309; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 280 to 286 in a biological sample.
312. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing symptoms and consequences of dysfunction, damage or disease of the gastrointestinal tract; said kit comprising:
- i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 280 or 282 to 286 in a sample of human DNA;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing symptoms and consequences of dysfunction, damage or disease of the gastrointestinal tract.

313. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing symptoms and consequences of dysfunction, damage or disease of the gastrointestinal tract; said kit comprising:
- i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core group of genes as defined in any of claims 281 to 286 in an expressed-protein-containing human sample;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing symptoms and consequences of dysfunction, damage or disease of the gastrointestinal tract.
314. A set of probes according to claim 280, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.
315. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to dysfunction, damage or disease of the respiratory system; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

#### KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME  
 T TRANSPORT & STORAGE  
 S STRUCTURAL  
 I IMMUNITY  
 N NERVOUS TRANSMISSION  
 G GROWTH & DIFFERENTIATION

RESPIRATORY GENE LIST	HUGO gene symbol	Protein function
11beta hydroxysteroid dehydrogenase 2	HSD11B2	E
2,3-bisphosphoglycerate mutase	BPGM	E
3 beta hydroxysteroid dehydrogenase 2	HSD3B2	E
Acetoacetyl 1-CoA-thiolase	ACAT1	E
Acetoacetyl 2-CoA-thiolase	ACAT2	E
Acetyl CoA synthase		E
Acetylcholine receptor, nicotinic, gamma	CHRNA7	N
Acetylcholinesterase	ACHE	E
Aconitase		E
Acyl CoA dehydrogenase, long chain	ACADL	E
Acyl CoA dehydrogenase, medium chain	ACADM	E
Acyl CoA dehydrogenase, short chain	ACADS	E
Acyl CoA dehydrogenase, very long chain	ACADVL	E

Adaptin, beta 3A	ADTB3A	T
Adenosine deaminase	ADA	E
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N
Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
Adrenocorticotrophic hormone (ACTH) receptor	ACTHR	G
Albumin, ALB	ALB	T
Alcohol dehydrogenase 1	ADH1	E
Alcohol dehydrogenase 2	ADH2	E
Alcohol dehydrogenase 3	ADH3	E
Alcohol dehydrogenase 4	ADH4	E
Alcohol dehydrogenase 5	ADH5	E
Alcohol dehydrogenase 6	ADH6	E
Alcohol dehydrogenase 7	ADH7	E
Aldolase A	ALDOA	E
Aldolase B	ALDOB	E
Aldolase C	ALDOC	E
Aldosterone receptor	MLR	G
Alpha 2 macroglobulin	A2M	I
Alpha1-antichymotrypsin	AACT	E
alpha1-antitrypsin	PI	E
alpha2-antiplasmin	PLI	E
alpha-actinin 2	ACTN2	G
alpha-actinin 3	ACTN3	G
alpha-Galactosidase A	GLA	E
alpha-ketoglutarate dehydrogenase		E
Aminopeptidase P	XPNPEP2	E
Amphiregulin	AREG	G
Androgen receptor	AR	G
Angiopoietin 1	ANGPT1	G
Angiopoietin 2	ANGPT2	G
Angiotensin converting enzyme	ACE, DCP1	E

Angiotensin receptor 1	AGTR1	T
Angiotensin receptor 2	AGTR2	T
Angiotensinogen	AGT	E
Annexin 1	ANX 1	I
Antidiuretic hormone receptor	ADHR	T
Antithrombin III	AT3	E
Apolipoprotein E	APOE	T
Arginase	ARG1	E
Arginine vasopressin	AVP	N
Arginine vasopressin receptor 1A	AVPR1A	N
Arginine vasopressin receptor 1B	AVPR1B	N
Arginine vasopressin receptor 2	AVPR2	N
Arginosuccinate lyase	ASL	E
Arylsulfatase D	ARSD	E
Arylsulfatase E	ARSE	E
Arylsulfatase F	ARSF	E
Aspartate transaminase		T
Ataxia telangiectasia gene, AT	ATM	G
ATP/ADP translocase		E
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G
Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
beta-galactosidase	GLB1	E
beta-Glucuronidase	GUSB	E
Biotinidase	BTD	E
Bloom syndrome protein	BLM	G
Bradykinin receptor B1		I
Bradykinin receptor B2		I
Butyrylcholinesterase	BCHE	E
C1 inhibitor		E
Cadherin E	CDH1	G
Cadherin EP		G
Cadherin N	CDH2	G
Cadherin P	CDH3	G
Calcitonin receptor /Calcitonin gene-related peptide receptor	CALCR	N
Calcitonin/Calcitonin gene-related peptide alpha	CALCA	N
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha-1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha-1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha-1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha-	CACNA1E	N

1E (CACNL1A6)		
Calcium channel, voltage-dependent, Alpha-2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N
Calcium channel, voltage-dependent, T-type		N
Calmodulin 1	CALM1	G
Calmodulin 2	CALM2	G
Calmodulin 3	CALM3	G
Calnexin	CANX	G
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Carnitine acetyltransferase	CRAT	E
Carnitine acylcarnitine translocase	CACT	E
Catalase	CAT	I
Cathepsin B		E
Cathepsin D		E
Cathepsin E		E
Cathepsin G	CTSG	E
Cathepsin H		E
Cathepsin K	CTSK	E
Cathepsin L		E
Cathepsin S		E
CD1	CD1	I
CD4	CD4	I
Cell adhesion molecule, intercellular, ICAM	ICAM1	G
Cell adhesion molecule, leukocyte-endothelial, LECAM (CD62)	LECAM1	G
Cell adhesion molecule, liver, LCAM	LCAM	G
Cell adhesion molecule, neural, NCAM1	NCAM1	G
Cell adhesion molecule, neural, NCAM120	NCAM120	G
Cell adhesion molecule, neural, NCAM2	NCAM2	G
Cell adhesion molecule, platelet-endothelial, PECAM	PECAM1	G
Cell adhesion molecule, vascular, VCAM	VCAM1	G
Chemokine receptor CXCR4	CXCR4	I
Chitotriosidase	chit	E
Cholecystokinin	CCK	N
Cholecystokinin B receptor	CCKBR	N
Choline acetyltransferase	CHAT	E
Citrate synthase		E
Coenzyme Q (CoQ)/ubiquinone		E
Collagen I alpha 1	COL1A1	S
Collagen I alpha 2	COL1A2	S

Collagen II alpha 1	COL2A1	S
Collagen III alpha 1	COL3A1	S
Collagen IV alpha 1	COL4A1	S
Collagen IV alpha 2	COL4A2	S
Collagen IV alpha 3	COL4A3	S
Collagen IV alpha 4	COL4A4	S
Collagen IV alpha 5	COL4A5	S
Collagen IV alpha 6	COL4A6	S
Collagen IX alpha 2	COL9A2, EDM2	S
Collagen IX alpha 3	COL9A3	S
Collagen receptor	COLR	S
Collagen V alpha 1	COL5A1	S
Collagen V alpha 2	COL5A2	S
Collagen VI alpha 1	COL6A1	S
Collagen VI alpha 2	COL6A2	S
Collagen VI alpha 3	COL6A3	S
Collagen VII alpha 1	COL7A1	S
Collagen X alpha 1	COL10A1	S
Collagen X alpha 1	COL11A1	S
Collagen XI alpha 2	COL11A2	S
Collagen XVII alpha 1	COL17A1	S
Colony-stimulating factor 1	CSF1	G
Colony-stimulating factor 1 receptor	CSF1R	G
Colony-stimulating factor 2	CSF2	G
Colony-stimulating factor 2 alpha receptor	CSF2RA	G
Colony-stimulating factor 2 beta receptor	CSF2RB	G
Colony-stimulating factor 3	CSF3	G
Colony-stimulating factor 3 receptor	CSF3R	G
Complement component C1 inhibitor	C1NH	I
Complement component C1qa	C1QA	I
Complement component C1qb	C1QB	I
Complement component C1qg	C1QG	I
Complement component C1r	C1R	I
Complement component C1s	C1S	I
Complement component C2	C2	I
Complement component C3	C3	I
Complement component C4A	C4A	I
Complement component C4B	C4B	I
Complement component C5	C5	I
Complement component C6	C6	I
Complement component C7	C7	I
Complement component C8	C8B	I
Complement component C9	C9	I
Complement component receptor 1	CR1	I
Complement component receptor 2	CR2	I
Complement component receptor 3	CR3	I
Complex I		E
Complex II		E

Complex III		E
Complex III		E
Complex V	MTATP6	E
Coproporphyrinogen oxidase	CPO	E
Corticotrophin-releasing hormone	CRH	T
Corticotrophin-releasing hormone receptor	CRHR1	T
Cortisol receptor		I
C-reactive protein CRP		I
Creatine kinase – B and m	CKBE	E
Creb binding protein	CREBBP	G
Cu <sup>2+</sup> transporting ATPase alpha polypeptide	ATP7A	E
Cyclic AMP-dependent protein kinase	PKA	E
Cyclic nucleotide phosphodiesterase 1B	PDE1B	E
Cyclic nucleotide phosphodiesterase 1B1	PDE1B1	E
Cyclic nucleotide phosphodiesterase 2A3	PDE2A3	E
Cyclic nucleotide phosphodiesterase 3A	PDE3A	E
Cyclic nucleotide phosphodiesterase 3B	PDE3B	E
Cyclic nucleotide phosphodiesterase 4A	PDE4A	E
Cyclic nucleotide phosphodiesterase 4C	PDE4C	E
Cyclic nucleotide phosphodiesterase 5A	PDE5A	E
Cyclic nucleotide phosphodiesterase 6A	PDE6A	E
Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E
Cyclic nucleotide phosphodiesterase 8	PDE8	E
Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclin-dependent kinase 2	CDK2	G
Cyclin-dependent kinase inhibitor 2A (p16)	CDKN2A	G
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
CYP11A1	CYP11A1	E
CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E
CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E
CYP2C18	CYP2C18	E



CYP2C19	CYP2C19	E
CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E
CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E
CYP2F1	CYP2F1	E
CYP2J2	CYP2J2	E
CYP3A3	CYP3A3	E
CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystathionase	CTH	E
Cystathione beta synthase	CBS	E
Cystic fibrosis transmembrane conductance regulator, CFTR	CFTR	N
Cytidine deaminase	CDA	E
Cytidine-5-prime-triphosphate synthetase	CTPS	E
Cytochrome a		E
Cytochrome b-245 alpha	CYBA	E
Cytochrome b-245 beta	CYBB	E
Cytochrome b-5	CYB5	E
Cytochrome c		E
Cytochrome c oxidase, MTCO		E
Cytokine-suppressive antiinflammatory drug- binding protein 1	CSBP1	I
Cytokine-suppressive antiinflammatory drug- binding protein 2	CSBP2	I
DAX1 nuclear receptor	DAX1	I
D-beta-hydroxybutyrate dehydrogenase		E
Delta 4-5 alpha-reductase		E
Desmin	DES	S
Dihydrolipoamide dehydrogenase	DLD	N
DNA glycosylases		E
Dopamine beta hydroxylase	DBH	E
Dopamine receptors D1	DRD1	N
Dopamine receptors D2	DRD2	N
Dopamine receptors D3	DRD3	N
Dopamine receptors D4	DRD4	N
Dopamine receptors D5	DRD5	N
Dystrophin	DMD	S

Elastase 1	ELAS1	E
Elastase 2	ELAS2	E
Elastin	ELN	S
Electron-transferring-flavoprotein alpha	ETFA	T
Electron-transferring-flavoprotein beta	ETFB	T
Electron-transferring flavoprotein dehydrogenase	ETFDH	E
Endothelin 1	EDN1	N
Endothelin 2	EDN2	N
Endothelin 3	EDN3	N
Endothelin converting enzyme	ECE1	N
Endothelin receptor type A	EDNRA	N
Endothelin receptor type B	EDNRB	N
Enolase	ENO1	E
Enoyl CoA hydratase		E
Enoyl CoA isomerase		E
Enoyl CoA reductase		E
Enterokinase	PRSS7, ENTK	E
Ephrin receptor tyrosine kinase A	EPHA	G
Ephrin receptor tyrosine kinase B	EPHB	G
Epidermal growth factor	EGF	G
Epidermal growth factor receptor	EGFR	G
Epoxide hydrolase 1, microsomal	EPHX1	E
Estrogen receptor	ESR	G
EWS RNA-binding protein	EWSR1	G
Eyes absent 1	EYA1	G
Faciogenital dysplasia	FGD1, FGDY	T
Factor 1 (No. one)	F1	I
Factor B, properdin		I
Factor D		I
Factor H	HF1	I
Factor I (letter I)	IF	I
Factor III	F3	I
Factor IX	F9	I
Factor V	F5	I
Factor VII	F7	I
Factor VIII	F8	I
Factor X	F10	I
Factor XI	F11	I
Factor XII	F12	I
Factor XIII A & B	F13A & F13B	I
Fc fragment of IgG, high affinity IA, receptor for	FCGR1A	G
Fc fragment of IgG, low affinity IIa, receptor for (CD32)	FCGR2A	G
Fc fragment of IgG, low affinity IIIa, receptor for (CD16)	FCGR3A	G
Fibrillin 1	FBN1	G

Fibrinogen alpha	FGA	S
Fibrinogen beta	FGB	S
Fibrinogen gamma	FGG	S
Fibroblast growth factor	FGF1	G
Fibroblast growth factor receptor 1	FGFR1	G
Fibroblast growth factor receptor 2	FGFR2	G
Fibroblast growth factor receptor 3	FGFR3	G
Fibronectin precursor	FN1	G
Flightless-II, Drosophila homolog of	FLII	G
Follicle stimulating hormone receptor	FSHR, ODG1	G
Follicle stimulating hormone, FSH	FSHB	G
Forkhead rhabdomyosarcoma gene	FKHR	G
Fructose-1,6-diphosphatase	FBP1	E
Furin		T
GABA receptor, alpha 1	GABRA1	N
GABA receptor, alpha 2	GABRA2	N
GABA receptor, alpha 3	GABRA3	N
GABA receptor, alpha 4	GABRA4	N
GABA receptor, alpha 5	GABRA5	N
GABA receptor, alpha 6	GABRA6	N
GABA receptor, beta 1	GABRB1	N
GABA receptor, beta 2	GABRB2	N
GABA receptor, beta 3	GABRB3	N
GABA receptor, gamma 1	GABRG1	N
GABA receptor, gamma 2	GABRG2	N
GABA receptor, gamma 3	GABRG3	N
GABA transaminase	ABAT	E
Galactocerebrosidase	GALC	E
Galactosyltransferase 1	GT1	G
Galactosyltransferase, alpha 1,3	GGTA1	G
Galactosyltransferase, beta 3	B3GALT	G
Glucocorticoid receptor	GRL	G
Glucokinase	GCK	E
Glucosidase, acid alpha	GAA	E
Glutamate dehydrogenase	GLUD1	E
Glutamate receptor 1	GLUR1	N
Glutamate receptor 2	GLUR2	N
Glutamate receptor 3	GLUR3	N
Glutamate receptor 4	GLUR4	N
Glutamate receptor 5	GLUR5	N
Glutamate receptor 6	GLUR6	N
Glutamate receptor 7	GLUR7	N
Glutamate receptor, ionotropic, NMDA 1	NMDAR1	N
Glutamate receptor, ionotropic, NMDA 2A	NMDAR2A	N
Glutamate receptor, ionotropic, NMDA 2B	NMDAR2B	N
Glutamate receptor, ionotropic, NMDA 2C	NMDAR2C	N
Glutamate receptor, ionotropic, NMDA 2D	NMDAR2D	N
Glutathione	GSH	T

Glutathione peroxidase, GPX1	GPX1	E
Glutathione peroxidase, GPX2	GPX2	E
Glutathione reductase, GSR	GSR	E
Glutathione S-transferase mu 1, GSTM1	GSTM1	E
Glutathione S-transferase mu 4, GSTM4		E
Glutathione S-transferase theta 1, GSTT1	GSTT1	E
Glutathione S-transferase theta 2, GSTT2		E
Glutathione S-transferase, GSTP1	GSTP1	E
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glutathione synthetase	GSS	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
Glycerol kinase	GK	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
GM2 ganglioside activator protein, GM2A	GM2A	E
Growth arrest-specific homeobox	GAX	G
Guanylyl cyclase		E
Haemoglobin alpha 1	HBA1	T
Haemoglobin alpha 2	HBA2	T
Haemoglobin beta	HBB	T
Haemoglobin delta	HBD	T
Haemoglobin gamma A	HBG1	T
Haemoglobin gamma B	HBG2	T
Haemoglobin gamma G	HBGG	T
Heat shock protein, HSP60		I
Heat shock protein, HSP70		I
Heat shock protein, HSP90		I
Heat shock protein, HSPA1		I
Heat shock protein, HSPA2		I
Heparin binding epidermal growth factor	HBEGF	G
Heparin Cofactor II	HCF2	I
Hermansky-pudlak syndrome gene	HPS	T
Hexokinase 1	HK1	E
Hexokinase 2	HK2	E
Hexosaminidase A	HEXA,TSD	E
Histamine receptors, H1		N
Histamine receptors, H2		N
Histamine receptors, H3		N
HMG-CoA lyase	HMGCL	E
HMG-CoA reductase	HMGCR	E
HMG-CoA synthase	HMGCS2	E
Holocarboxylase synthetase	HLCS	E
Hyaluronidase		T
Hypoxia inducible factor 1	HIF1A	E
Hypoxia inducible factor 2		E
Immunoglobulin E (IgE) reponsiveness gene	IGER	I
Immunoglobulin E (IgE) serum concentration	IGES	I

regulator gene		
Immunoglobulin gamma (IgG) 2	IGHG2	I
Insulin	INS	G
Insulin receptor	INSR	G
Insulin-like growth factor 1	IGF1	G
Insulin-like growth factor 1 receptor	IGF1R	G
Insulin-like growth factor 2	IGF2	G
Insulin-like growth factor 2 receptor	IGF2R	G
Integrin beta 1	ITGB1	G
Integrin beta 2	ITGB2	G
Integrin beta 5	ITGB5	G
Integrin beta 6	ITGB6	G
Integrin, alpha M	ITGAM	G
Inter-alpha-trypsin inhibitor, IAT1		E
Interferon alpha	IFNA1	I
Interferon beta	IFNB	I
Interferon gamma	IFNG	I
Interferon gamma receptor 1	IFNGR1	I
Interferon gamma receptor 2	IFNGR2	I
Interferon regulatory factor 1	IRF1	I
Interferon regulatory factor 4	IRF4	I
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) 10	IL10	I
Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I
Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I

Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
Isocitrate dehydrogenase		E
Kallikrein 3	KAK3	I
Kininogen, High molecular weight	KNG	I
Kynureninase		E
Laminin 5, alpha 3	LAMA3	G
Laminin 5, beta 3	LAMB3	G
Laminin 5, gamma 2	LAMC2	G
Laminin M	LAMM	G
Laminin receptor 1	LAMR1	G
Latent transforming growth factor-beta binding protein 2	LTBP2	G
Lecithin-cholesterol acyltransferase	LCAT	E
Leptin	LEP	G
Leptin receptor	LEPR	G
Leukotriene A4 hydrolase		I
Leukotriene A4 synthase	LTA4S	E
Leukotriene B4 receptor		I
Leukotriene B4 synthase	LTB4S	E
Leukotriene C4 receptor		I
Leukotriene C4 synthase	LTC4S	E
Leukotriene D4/E4 receptor		I
LIM homeobox protein 1	LHX1	G
Lipoamide dehydrogenase	OGDH	E
Lipoprotein lipase	LPL	I
Lipoprotein receptor, Low Density	LDLR	T
Lipoprotein, High Density	HDLDT1	T
Lipoprotein, Intermediate Density		T
Lipoprotein, Low Density 1		T
Lipoprotein, Low Density 2		T
Lipoprotein, Very Low Density	VLDLR	T
Lipoxygenase		E
Low density lipoprotein receptor-related protein precursor	LRP	T
Lymphoid enhancer-binding factor	LEF-1	G
Lysosomal acid lipase	LIPA	E
Lysozyme	LYZ	I
MAD (mothers against decapentaplegic, Drosophila) homologue 4	MADH4	G
Malate dehydrogenase, mitochondrial	MDH2	E
Malonyl CoA transferase		E
Mannose binding protein	MBP	I
Mannosidase, alpha B lysosomal	MANB	E
Mannosidase, beta A lysosomal	MANBA	E
Matrix Gla protein	MGP	G
Matrix metalloproteinase 1	MMP1	E
Matrix metalloproteinase 10	MMP10	E

Matrix metalloproteinase 11	MMP11	E
Matrix metalloproteinase 12	MMP12	E
Matrix metalloproteinase 13	MMP13	E
Matrix metalloproteinase 14	MMP14	E
Matrix metalloproteinase 15	MMP15	E
Matrix metalloproteinase 16	MMP16	E
Matrix metalloproteinase 17	MMP17	E
Matrix metalloproteinase 18	MMP18	E
Matrix metalloproteinase 19	MMP19	E
Matrix metalloproteinase 2	MMP2	E
Matrix metalloproteinase 3	MMP3, STMY1	E
Matrix metalloproteinase 4	MMP4	E
Matrix metalloproteinase 5	MMP5	E
Matrix metalloproteinase 6	MMP6	E
Matrix metalloproteinase 7	MMP7	E
Matrix metalloproteinase 8	MMP8	E
Matrix metalloproteinase 9	MMP9	E
Methionine adenosyltransferase	MAT1A, MAT2A	E
Midline 1	MID1	G
Mitochondrial trifunctional protein, alpha subunit	HADHA	E
Mitochondrial trifunctional protein, beta subunit	HADHB	E
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Myoglobin		T
Myotubularin	MTM1	S
Na <sup>+</sup> , K <sup>+</sup> ATPase, alpha	ATP1A1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 1	ATP1B1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 2	ATP1B2	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 3	ATP1B3	G
NADH dehydrogenase		E
NADH dehydrogenase (ubiquinone) Fe-S protein 1	NDUFS1	E
NADH dehydrogenase (ubiquinone) Fe-S protein 4	NDUFS4	E
NADH dehydrogenase (ubiquinone) flavoprotein 1	NDUFV1	E
NADH-cytochrome b5 reductase	DIA1	E
NADPH-dependent cytochrome P450 reductase	POR	E
Nebulin	NEB	S
Nephrosis 1	NPHS1	T

Nerve growth factor	NGF	G
Nerve growth factor receptor	NGFR	G
Neuraminidase sialidase	NEU	T
Neuregulin	HGL	G
Neurofibromin 1	NF1	G
Neurofibromin 2	NF2	G
Neurokinin A	NKNA	N
Neurokinin B	NKNB	N
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Nitric oxide synthase 1, NOS1	NOS1	E
Nitric oxide synthase 2, NOS2	NOS2	E
Nitric oxide synthase 3, NOS3	NOS3	E
Notch ligand - jagged 1	JAG1, AGS	G
Nucleoside diphosphate kinase-A	NDPKA	E
Oncogene ELK1	ELK1	G
Oncogene ELK2	ELK2	G
Oncogene sis	PDGFB	G
Ornithine delta-aminotransferase	OAT	E
Paired box homeotic gene 6	PAX6	G
Parathyroid hormone	PTH	G
Parathyroid hormone receptor	PTHr1	G
Parathyroid hormone related-peptide	PTHrP	G
Parathyroid hormone-like hormone	PTHrLH	G
Patched (Drosophila) homolog, PTCH	PTCH	G
Peroxisomal membrane protein 3	PXMP3	T
Peroxisome biogenesis factor 1	PEX1	T
Peroxisome biogenesis factor 19	PEX19	T
Peroxisome biogenesis factor 6	PEX6	T
Peroxisome biogenesis factor 7	PEX7	T
Peroxisome receptor 1	PXR1	T
Phenylalanine hydroxylase	PAH	E
Phenylalanine monooxygenase		E
Phenylethanolamine N-methyltransferase, PNMT	PNMT	E
Phosphofructokinase, liver	PFKL	E
Phosphofructokinase, muscle	PFKM	E
Phosphoglucomutase		E
Phosphoglucose isomerase	GPI	E
Phosphoglycerate kinase 1	PGK1	E
Phosphoglycerate mutase 2	PGAM2	E
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I



Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C epsilon		I
Pineolytic beta-receptors		E
Plasminogen	PLG	E
Plasminogen activator inhibitor 1	PAI1	E
Plasminogen activator inhibitor 2	PAI2	E
Plasminogen activator receptor, Urokinase	UPAR; PLAUR	S
Plasminogen activator, Tissue	PLAT; TPA	E
Plasminogen activator, Urokinase	UPA; PLAU	E
Platelet derived growth factor	PDGF	G
Platelet derived growth factor receptor	PDGFR	G
Platelet-activating factor receptor	PAFR	I
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium voltage-gated channel E1	KCNE1	N
Prekallikrein		I
Procollagen N-protease		E
Progesterone receptor (RU486 binding receptor)	PGR	G
Proliferin	PLF	G
Proopiomelanocortin	POMC	N
Properdin P factor, complement	PFC, PFD	I
Prosaposin	PSAP	N
Prostacyclin synthase		I
Prostaglandin 15-OH dehydrogenase	HGPD; PGDH	I
Prostaglandin D - DP receptor		I
Prostaglandin E1 receptor		I
Prostaglandin E2 receptor		I
Prostaglandin E3 receptor		I
Prostaglandin F - FP receptor		I
Prostaglandin F2 alpha receptor		I
Prostaglandin I2 receptor		T
Prostaglandin IP receptor		I
Protein C	PROC	I
Protein C inhibitor	PCI	I
Protein phosphatase 2, regulatory subunit A, beta isoform	PPP2R1B	E
Protein S	PROS1	I
Prothrombin precursor	F2	I
Pyruvate carboxylase	PC	E
Pyruvate decarboxylase	PDHA	E
Pyruvate kinase	PKLR	E
Quinoid dihydropteridine reductase	QDPR	E
Renin	REN	E
Replication factor C	RFC2	E
Retinoblastoma 1	RB1	G
RIGUI	RIGUI	G
Salivary amylase, AMY1		T

Selectin E	SELE	N
Selectin L	SELL	N
Selectin P	SELP	N
Serine hydroxymethyltransferase	SHMT	E
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage gated, type IV, alpha polypeptide	SCN4A	N
Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Solute carrier family 21, member 2	SLC21A2	T
Solute carrier family 4 (anion exchanger), member 1	SLC4A1	T
Solute carrier family 4 (anion exchanger), member 2	SLC4A2	T
Solute carrier family 4 (anion exchanger), member 3	SLC4A3	T
Solute carrier family 6 (GAMMA-AMINOBUTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Somatostatin receptor, SSTR2	SSTR2	G
Sphingomyelinase	SMPD1	E
Substance P		N
Succinate dehydrogenase 2	SDH2	E
Succinate thiokinase		E
Succinyl CoA synthase		E
Superoxide dismutase 1	SOD1	E
Superoxide dismutase 3	SOD3	E

Surfactant pulmonary-associated protein A1	SFTPA1	T
Surfactant pulmonary-associated protein A2	SFTPA2	T
Surfactant pulmonary-associated protein B	SFTPB	T
Surfactant pulmonary-associated protein C	SFTPC	T
Surfactant pulmonary-associated protein D	SFTPD	T
Surfeit 1	SURF1	G
Survival of motor neuron 1, telomeric	SMN1	T
Talin	TLN	G
T-BOX 2	TBX2	G
T-BOX 3	TBX3	G
TEK, tyrosine kinase, endothelial	TEK	E
Telomerase protein component		E
Thiolase, peroxisomal		E
Thrombin receptor	F2R	I
Thrombomodulin	THBD	I
Thrombopoietin	THPO	G
Thrombospondin	THBS1	G
Thromboxane A synthase 1	TBXAS1	I
Thromboxane A2	TXA2	I
Thromboxane A2 receptor	TBXA2R	I
Thyroglobulin	TG	G
Thyroid hormone receptor, alpha	THRA	G
Thyroid hormone receptor, beta	THRB	G
Thyroid peroxidase	TPO	G
Thyroid receptor auxiliary protein	TRAP	G
Thyroid-stimulating hormone receptor	TSHR	G
Thyroid-stimulating hormone, alpha	TSHA	G
Thyroid-stimulating hormone, beta	TSHB	G
Thyrotropin releasing hormone receptor	TRHR	G
Topoisomerase I		E
Transacylase		E
Transferrin	TF	G
Transferrin receptor	TFRC	G
Transforming growth factor, beta 2	TGFB2	G
Transforming growth factor, beta induced	TGFB1	G
Transforming growth factor, beta receptor 2	TGFBR2	G
Transketolase	TKT	E
Transketolase-like 1	TKTL1	E
Triosephosphate isomerase	TPI1	E
Trypsin inhibitor		E
Uncoupling protein 1		T
Uroporphyrinogen III synthase	UROS	E
Vasoactive intestinal polypeptide receptor	VIPR	N
Vasoinhibitory peptide		G
Vitronectin receptor, alpha	VNRA	T
Von Hippel-Lindau gene	VHL	G
Wolf-Hirschhorn syndrome candidate 1 gene	WHSC1	G
Xanthine dehydrogenase	XDH	E

316. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 315.
317. A set according to claim 315 or 316 in which a minority of said probes for listed genes are absent.
318. A set according to claim 315 or 316 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.
319. A set according to claim 315 or 316 in which a limited number of probes are replaced by probes for non-listed genes.
320. A set of probes for a core group of genes according to any of claims 315 to 319 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.
321. A set according to any of claims 315 to 320 consisting of probes for members of a sub-group of the core group.
322. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.
323. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.
324. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.
325. A set according to claim 322 or 323 in which said substrate is a semiconductor microchip.
326. A set according to any preceding claim for use in a biological assay for detection of said gene variants.
327. A set according to any preceding claim for use in the measurement of differential gene expression levels.
328. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
329. A medical device including a set according to any of claims 315 to 327 for use in an array for detection of differential gene expression levels.
330. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 315) in a target group of genes by hybridising a nucleic acid-containing

- sample from said patient or individual to a set according to any of claims 315 and 317 to 327 and relating the probe hybridisation pattern to said variations.
331. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 316) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 316 to 327 and relating the probe interaction pattern to said variations.
332. Use of a set or device according to any of claims 315 to 327 for the prognosis and management of patients suffering from or at risk of dysfunction, damage or disease of the respiratory system or experiencing the clinical or social consequences following dysfunction, damage or disease of the respiratory system.
333. Use of a set or device according to any of claims 315 to 327 for predicting likely therapeutic response and adverse events following therapeutic intervention.
334. Use of a set or device according to any of claims 315 to 327 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
335. Use of a set or device according to any of claims 315 to 327 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
336. Use of a set or device according to any of claims 315 to 327 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
337. Use of a set or device according to any of claims 315 to 327 for the development of new strategies of therapeutic intervention and in clinical trials.
338. Use of a set or device according to any of claims 315 to 327 for construction of and generation of algorithms for patient and healthcare management.
339. Use of a set or device according to any of claims 315 to 327 for modelling or assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations
340. Use of a set or device according to any of claims 315 to 327 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
341. Use of a set or device according to any of claims 315 to 327 for predicting optimum configuration/management of thereapeutic intervention.
342. A method according to claim 330 or 331 in which the identification of gene variants is indicative of a higher risk of developing the symptoms of, and/or the clinical or social consequences following, dysfunction, damage or disease of the respiratory system for the patient or individual.
343. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop the symptoms of, and/or the clinical or social consequences following, dysfunction, damage or disease of the respiratory system, which method comprises:
- i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from the symptoms of, and/or the clinical or social consequences following, dysfunction, damage or disease of the respiratory system:

- ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the symptoms of, and/or the clinical or social consequences following, dysfunction, damage or disease of the respiratory system;
  - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 315 to 321;
  - iv) calculating the frequencies of these alleles in the samples from i) and ii);
  - v) comparing the frequencies of these alleles in i) and ii);
  - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing the symptoms of, and/or the clinical or social consequences following, dysfunction, damage or disease of the respiratory system.
344. A method for assessing whether a given subject will be at risk of developing symptoms, which comprises comparing said subject's genotype with a model generated by the method of claim 343.
345. A method according to any of claims 330, 331, 343 and 344 wherein at least one step is computer-controlled.
346. An assay suitable for use in a method according to any of claims 330, 331, 343 and 344; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 315 to 321 in a biological sample.
347. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing the symptoms of, and/or the clinical or social consequences following, dysfunction, damage or disease of the respiratory system; said kit comprising:
- i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 315 or 317 to 321 in a sample of human DNA;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing the symptoms of, and/or the clinical or social consequences following, dysfunction, damage or disease of the respiratory system.
348. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing the symptoms of, and/or the clinical or social consequences following, dysfunction, damage or disease of the respiratory system; said kit comprising:
- i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core group of genes as defined in any of claims 316 to 321 in an expressed-protein-containing human sample;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing the symptoms of, and/or the clinical or social consequences following, dysfunction, damage or disease of the respiratory system.
349. A set of probes according to claim 315, wherein the probes are selected from

the group consisting of oligonucleotides and polynucleotides.

350. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to injury, inflammation, infection, immunity and/or repair; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

#### KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME  
T TRANSPORT & STORAGE  
S STRUCTURAL  
I IMMUNITY  
N NERVOUS TRANSMISSION  
G GROWTH & DIFFERENTIATION

IMMUNITY GENE LIST	HUGO gene symbol	Protein function
5,10-methylenetetrahydrofolate reductase (NADPH)	MTHFR	E
Acetylcholinesterase	ACHE	E
Acidic amino acid transporter		T
Actin, alpha, cardiac	ACTC	S
Actin, alpha, skeletal	ACTA1	S
Actin, alpha, smooth, aortic	ACTA2	S
Actin, beta	ACTB	S
Actin, gamma 2	ACTG2	S
ADAM (A disintegrin and metalloproteinase) 1	ADAM1	E
ADAM (A disintegrin and metalloproteinase) 10	ADAM10	E
ADAM (A disintegrin and metalloproteinase) 11	ADAM11	E
ADAM (A disintegrin and metalloproteinase) 12	ADAM12	E
ADAM (A disintegrin and metalloproteinase) 13	ADAM13	E
ADAM (A disintegrin and metalloproteinase) 14	ADAM14	E
ADAM (A disintegrin and metalloproteinase) 15	ADAM15	E
ADAM (A disintegrin and metalloproteinase) 16	ADAM16	E
ADAM (A disintegrin and metalloproteinase) 17	ADAM17	E
ADAM (A disintegrin and metalloproteinase) 18	ADAM18	E
ADAM (A disintegrin and metalloproteinase) 19	ADAM19	E
ADAM (A disintegrin and metalloproteinase) 2	ADAM2	E
ADAM (A disintegrin and metalloproteinase)	ADAM3A	E
3A		
ADAM (A disintegrin and metalloproteinase)	ADAM3B	E
3B		
ADAM (A disintegrin and metalloproteinase) 4	ADAM4	E

ADAM (A disintegrin and metalloproteinase) 5	ADAM5	E
ADAM (A disintegrin and metalloproteinase) 6	ADAM6	E
ADAM (A disintegrin and metalloproteinase) 7	ADAM7	E
ADAM (A disintegrin and metalloproteinase) 8	ADAM8	E
ADAM (A disintegrin and metalloproteinase) 9	ADAM9	E
Adducin, alpha	ADD1	S
Adducin, beta	ADD2	S
Adenosine deaminase	ADA	E
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N
Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
Adrenocorticotrophic hormone (ACTH) receptor	ACTHR	G
Albumin, ALB	ALB	T
Aldosterone receptor	MLR	G
Alpha 1 acid glycoprotein	AAG; AGP	T
Alpha 2 macroglobulin	A2M	I
alpha1-antitrypsin	PI	E
alpha2-antiplasmin	PLI	E
Alpha-fetoprotein	AFP	G
alpha-glucosidase, neutral AB	GANAB	E
alpha-glucosidase, neutral C	GANC	E
Aminopeptidase P	XPNPEP2	E
Amylo-1,6-glucosidase	AGL	E
Amyloid beta A4 precursor protein	APP	N
Amyloid beta A4 precursor-like protein	APLP	N
Androgen binding protein	ABP	T
Androgen receptor	AR	G
Angiopoietin 1	ANGPT1	G
Angiopoietin 2	ANGPT2	G
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensin receptor 1	AGTR1	T
Angiotensin receptor 2	AGTR2	T



Angiotensinogen	AGT	E
Annexin 1	ANX 1	I
Antidiuretic hormone receptor	ADHR	T
Anti-Mullerian hormone	AMH	G
Antithrombin III	AT3	E
Apaf-1		S
Apolipoprotein E	APOE	T
Apoptosis antigen 1	APT1	I
Apoptosis antigen ligand 1	APT1LG1	I
Apoptosis-inducing factor	AIF	I
Arginosuccinate lyase	ASL	E
Aryl hydrocarbon receptor	AHR	T
Asparagine synthetase	AS	E
Aspartylglucosaminidase	AGA	E
Ataxia telangiectasia complementation group D	ATD, ATDC	G
Ataxia telangiectasia gene, AT	ATM	G
ATP-binding cassette transporter 7	ABC7	I
Attractin		I
Autoimmune regulator, AIRE	AIRE	I
B-cell CLL/lymphoma 1	BCL1	I
B-cell CLL/lymphoma 10	BCL10	I
B-cell CLL/lymphoma 3	BCL3	I
B-cell CLL/lymphoma 4	BCL4	I
B-cell CLL/lymphoma 5	BCL5	I
B-cell CLL/lymphoma 6	BCL6	I
B-cell CLL/lymphoma 7	BCL7	I
B-cell CLL/lymphoma 8	BCL8	I
B-cell CLL/lymphoma 9	BCL9	I
BCL2-associated X protein	BAX	G
BCL2-related protein A1	BCL2A1	G
Beckwith-Wiedemann region 1A	BWR1A	G
beta 2 microglobulin	B2M	I
Bleomycin hydrolase	BLMH	E
Bloom syndrome protein	BLM	G
Bradykinin receptor B1		I
Bradykinin receptor B2		I
Brain derived neurotrophic factor	BDNF	G
Brain derived neurotrophic factor (BDNF) receptor	BDNFR	G
BRCA1-associated RING domain gene 1	BARD1	G
Breakpoint cluster region	BCR	G
Breast cancer 1	BRCA1	G
Breast cancer 2	BRCA2	G
Breast cancer, ductal, 1	BRCD1	G
Breast cancer, ductal, 2	BRCD2	G
Butyrylcholinesterase	BCHE	E
C3 convertase		E
Cadherin E	CDH1	G

Cadherin EP		G
Cadherin N	CDH2	G
Cadherin P	CDH3	G
Calbindin 1	CALB1	G
Calbindin D9K	CALB3	G
Calcineurin A1	CALNA1	I
Calcineurin A2	CALNA2	I
Calcineurin A3	CALNA3	I
Calcineurin B		I
Calcitonin receptor /Calcitonin gene-related peptide receptor	CALCR	N
Calcitonin/Calcitonin gene-related peptide alpha	CALCA	N
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha-1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha-1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha-1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha-1E (CACNL1A6)	CACNA1E	N
Calcium channel, voltage-dependent, Alpha-2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, L type, alpha 1S subunit	CACNA1S	N
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N
Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	CACNA1A	N
Calcium channel, voltage-dependent, T-type		N
Calmodulin 1	CALM1	G
Calmodulin 2	CALM2	G
Calmodulin 3	CALM3	G
Calmodulin-dependant protein kinase II	CAMK2A	G
Calnexin	CANX	G
Calpain	CAPN, CAPN3	E
Calretinin	CALB2	N
Canalicular multispecific organic anion transporter	CMOAT	T
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Carboxylesterase 1	CES1	E

Cardiac-specific homeobox, CSX	CSX	G
Cartilage-hair hypoplasia gene	CHH	N
Caspase 1	CASP1	G
Catalase	CAT	I
Cathepsin G	CTSG	E
CD1	CD1	I
CD10	CD10	I
CD100	CD100	I
CD101	CD101	I
CD103	CD103	I
CD106	CD106	I
CD107	CD107	I
CD108	CD108	I
CD109	CD109	I
CD110	CD110	I
CD111	CD111	I
CD112	CD112	I
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CD117	CD117	I
CD118	CD118	I
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CD124	CD124	I
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CD91	CD91	
CD92	CD92	

CD93	CD93	I
CD94	CD94	I
CD96	CD96	I
CD97	CD97	I
CD98	CD98	I
CD99	CD99	I
Cell adhesion molecule, intercellular, ICAM	ICAM1	G
Cell adhesion molecule, leukocyte-endothelial, LECAM (CD62)	LECAM1	G
Cell adhesion molecule, liver, LCAM	LCAM	G
Cell adhesion molecule, neural, NCAM1	NCAM1	G
Cell adhesion molecule, neural, NCAM120	NCAM120	G
Cell adhesion molecule, neural, NCAM2	NCAM2	G
Cell adhesion molecule, platelet-endothelial, PECAM	PECAM1	G
Cell adhesion molecule, vascular, VCAM	VCAM1	G
Chediak-Higashi syndrome 1 gene	CHS1	T
Chemokine MCAF	MCAF	I
Chemokine receptor CCR2	CCR2	I
Chemokine receptor CCR3	CCR3	I
Chemokine receptor CCR5	CCR5	I
Chemokine receptor CXCR1	CXCR1	I
Chemokine receptor CXCR2	CXCR2	I
Chemokine receptor CXCR4	CXCR4	I
Cholesterylester hydrolase		I
Chondritin Sulphate A - placental receptor		I
Chromogranin A	CHGA	G
Chymase	CHY1	
Clathrin		T
CoA transferase		E
Collagen I alpha 1	COL1A1	S
Collagen I alpha 2	COL1A2	S
Collagen II alpha 1	COL2A1	S
Collagen III alpha 1	COL3A1	S
Collagen IV alpha 1	COL4A1	S
Collagen IV alpha 2	COL4A2	S
Collagen IV alpha 3	COL4A3	S
Collagen IV alpha 4	COL4A4	S
Collagen IV alpha 5	COL4A5	S
Collagen IV alpha 6	COL4A6	S
Collagen IX alpha 2	COL9A2, EDM2	S
Collagen IX alpha 3	COL9A3	S
Collagen receptor	COLR	S
Collagen V alpha 1	COL5A1	S
Collagen V alpha 2	COL5A2	S
Collagen VI alpha 1	COL6A1	S
Collagen VI alpha 2	COL6A2	S
Collagen VI alpha 3	COL6A3	S

Collagen VII alpha 1	COL7A1	S
Collagen X alpha 1	COL10A1	S
Collagen X alpha 1	COL11A1	S
Collagen XI alpha 2	COL11A2	S
Collagen XVII alpha 1	COL17A1	S
Collagenic-like tail subunit of asymmetric acetylcholinesterase	COLQ	E
Colony-stimulating factor 1	CSF1	G
Colony-stimulating factor 1 receptor	CSF1R	G
Colony-stimulating factor 2	CSF2	G
Colony-stimulating factor 2 alpha receptor	CSF2RA	G
Colony-stimulating factor 2 beta receptor	CSF2RB	G
Colony-stimulating factor 3	CSF3	G
Colony-stimulating factor 3 receptor	CSF3R	G
Complement component C1 inhibitor	C1NH	I
Complement component C1qa	C1QA	I
Complement component C1qb	C1QB	I
Complement component C1qg	C1QG	I
Complement component C1r	C1R	I
Complement component C1s	C1S	I
Complement component C2	C2	I
Complement component C3	C3	I
Complement component C4A	C4A	I
Complement component C4B	C4B	I
Complement component C5	C5	I
Complement component C6	C6	I
Complement component C7	C7	I
Complement component C8	C8B	I
Complement component C9	C9	I
Complement component receptor 1	CR1	I
Complement component receptor 2	CR2	I
Complement component receptor 3	CR3	I
Contactin	CNTN1	G
Core-binding factor, alpha 1	CBFA1	G
Core-binding factor, alpha 2	CBFA2	G
Core-binding factor, beta	CBFB	G
Cortico-steroid binding protein		T
Corticosteroid nuclear receptor		I
Corticotrophin-releasing hormone	CRH	T
Corticotrophin-releasing hormone receptor	CRHR1	T
Cortisol receptor		I
C-reactive protein CRP		I
c-src tyrosine kinase	CSK	G
Cyclic AMP response element binding protein	CREB	G
Cyclic AMP-dependent protein kinase	PKA	E
Cyclic nucleotide phosphodiesterase 1B	PDE1B	E
Cyclic nucleotide phosphodiesterase 1B1	PDE1B1	E
Cyclic nucleotide phosphodiesterase 2A3	PDE2A3	E

Cyclic nucleotide phosphodiesterase 3A	PDE3A	E
Cyclic nucleotide phosphodiesterase 3B	PDE3B	E
Cyclic nucleotide phosphodiesterase 4A	PDE4A	E
Cyclic nucleotide phosphodiesterase 4C	PDE4C	E
Cyclic nucleotide phosphodiesterase 5A	PDE5A	E
Cyclic nucleotide phosphodiesterase 6A	PDE6A	E
Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E
Cyclic nucleotide phosphodiesterase 8	PDE8	E
Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclin D	CCND1	G
Cyclin-dependent kinase 1	CDK1	G
Cyclin-dependent kinase 10	CDK10	G
Cyclin-dependent kinase 2	CDK2	G
Cyclin-dependent kinase 3	CDK3	G
Cyclin-dependent kinase 4	CDK4	G
Cyclin-dependent kinase 5	CDK5	G
Cyclin-dependent kinase 6	CDK6	G
Cyclin-dependent kinase 7	CDK7	G
Cyclin-dependent kinase 8	CDK8	G
Cyclin-dependent kinase 9	CDK9	G
Cyclin-dependent kinase inhibitor 1A (P21, CIP1)	CDKN1A	G
Cyclin-dependent kinase inhibitor 1B (P27, KIP1)	CDKN1B	G
Cyclin-dependent kinase inhibitor 1C (P57, KIP2)	CDKN1C	G
Cyclin-dependent kinase inhibitor 2A (p16)	CDKN2A	G
Cyclin-dependent kinase inhibitor 3	CDKN3	G
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
Cyclophilin		I
CYP11A1	CYP11A1	E
CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E



CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E
CYP2C18	CYP2C18	E
CYP2C19	CYP2C19	E
CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E
CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E
CYP2F1	CYP2F1	E
CYP2J2	CYP2J2	E
CYP3A3	CYP3A3	E
CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystathionase	CTH	E
Cystathione beta synthase	CBS	E
Cystic fibrosis transmembrane conductance regulator, CFTR	CFTR	N
Cytidine deaminase	CDA	E
Cytidine-5-prime-triphosphate synthetase	CTPS	E
Cytochrome a		E
Cytochrome c		E
Cytochrome c oxidase, MTCO		E
Cytokine-suppressive antiinflammatory drug- binding protein 1	CSBP1	I
Cytokine-suppressive antiinflammatory drug- binding protein 2	CSBP2	I
Defender against cell death 1	DAD1	G
Deleted in colorectal carcinoma	DCC	G
Deoxycorticosterone (DOC) receptor		E
Deoxycytidine kinase DCK		E
Dihydrolipoyl dehydrogenase 2	PDHA	E
Dihydrolipoyl transacetylase	PDHA	E
Dopamine receptors D1	DRD1	N
Dopamine receptors D2	DRD2	N
Dopamine receptors D3	DRD3	N
Dopamine receptors D4	DRD4	N
Dopamine receptors D5	DRD5	N
Duffy blood group	FY	T
Dynamin	DNM1	G

EB1		G
Elastase 1	ELAS1	E
Elastase 2	ELAS2	E
Endoglin	ENG	S
Endo-P-D-glucuronidase		I
Enolase	ENO1	E
Erythroid kruppel-like factor	EKLF	G
Erythropoietin	EPO	I
Erythropoietin receptor	EPOR	I
Estrogen receptor	ESR	G
EWS RNA-binding protein	EWSR1	G
Factor 1 (No. one)	F1	I
Factor B, properdin		I
Factor D		I
Factor H	HF1	I
Factor I (letter I)	IF	I
Factor III	F3	I
Factor IX	F9	I
Factor V	F5	I
Factor VII	F7	I
Factor VIII	F8	I
Factor X	F10	I
Factor XI	F11	I
Factor XII	F12	I
Factor XIII A & B	F13A & F13B	I
Fanconi anemia, complementation group C	FANCC	T
Fanconi anemia, complementation group D	FANCD	T
Fc fragment of IgG, low affinity IIa, receptor for (CD32)	FCGR2A	G
Fc receptor		I
Fibrinogen alpha	FGA	S
Fibrinogen beta	FGB	S
Fibrinogen gamma	FGG	S
Fibronectin precursor	FN1	G
Follicle stimulating hormone receptor	FSHR, ODG1	G
Follicle stimulating hormone, FSH	FSHB	G
Follicular lymphoma variant translocation 1	FVT1	I
Forkhead rhabdomyosarcoma gene	FKHR	G
Forkhead transcription factor 7	FKHL7	G
Galactosyltransferase 1	GT1	G
Galactosyltransferase, alpha 1,3	GGTA1	G
Galactosyltransferase, beta 3	B3GALT	G
Glial-cell derived neurotrophic factor (GDNF) receptor		N
Glial-cell derived neurotrophic factor, GDNF	GDNF	N
Glucosaminyl (N-acetyl) transferase 2, I-branching enzyme	GCNT2	E
Glutamate receptor 1	GLUR1	N

Glutamate receptor 2	GLUR2	N
Glutamate receptor 3	GLUR3	N
Glutamate receptor 4	GLUR4	N
Glutamate receptor 5	GLUR5	N
Glutamate receptor 6	GLUR6	N
Glutamate receptor 7	GLUR7	N
Glutamate receptor, ionotropic, NMDA 1	NMDAR1	N
Glutamate receptor, ionotropic, NMDA 2A	NMDAR2A	N
Glutamate receptor, ionotropic, NMDA 2B	NMDAR2B	N
Glutamate receptor, ionotropic, NMDA 2C	NMDAR2C	N
Glutamate receptor, ionotropic, NMDA 2D	NMDAR2D	N
Glutamine synthase		E
Glutathione	GSH	T
Glutathione peroxidase, GPX1	GPX1	E
Glutathione peroxidase, GPX2	GPX2	E
Glutathione S-transferase mu 1, GSTM1	GSTM1	E
Glutathione S-transferase mu 4, GSTM4		E
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
Glycerol kinase	GK	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
Glycophorin A	GYP A	S
Glycophorin B	GYPB	S
Glycophorin C	GYP C	S
Glycosyltransferases, ABO blood group	ABO	E
Glypican 3	GPC3, SDYS	G
Gonadotropin releasing hormone receptor	GNRHR	G
Growth-regulated protein precursor, GRO	GRO	I
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 1, GNAI1	GNAI1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 2, GNAI2	GNAI2	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 3, GNAI3	GNAI3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS1	GNAS1	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS2	GNAS2	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS3	GNAS3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS4	GNAS4	N
Guanine nucleotide-binding protein, q polypeptide	GNAQ	N
H(+), K(+) - ATPase	ATP4B	N
Haemoglobin alpha 1	HBA1	T

Haemoglobin alpha 2	HBA2	T
Haemoglobin beta	HBB	T
Haemoglobin delta	HBD	T
Haemoglobin gamma A	HBG1	T
Haemoglobin gamma B	HBG2	T
Haemoglobin gamma G	HBGG	T
Haptoglobin, alpha 1	HPA1	I
Haptoglobin, alpha 2	HPA2	I
Haptoglobin, beta	HPB	I
Hemochromatosis	HFE	T
Heparin binding epidermal growth factor	HBEGF	G
Heparin Cofactor II	HCF2	I
Hepatitis B virus integration site 1	HVBS1	I
Hepatitis B virus integration site 2	HVBS6	I
High mobility group protein C	HMGIC	G
High mobility group protein Y	HMG1Y	G
Histamine receptors, H1		N
Histamine receptors, H2		N
Histamine receptors, H3		N
Histatin 1		I
Histatin 2		I
Histatin 3	HTN3	I
HLA-B associated transcript 1	BAT1	I
Holocarboxylase synthetase	HLCS	E
Homeobox 11	HOX11	G
Homeobox HB24	HLX1	G
IC7 A and B		I
Ikaros gene	IKAROS	G
Immunoglobulin alpha (IgA)	IGHA	I
Immunoglobulin delta (IgD)	IGHD	I
Immunoglobulin E (IgE) responsiveness gene	IGER	I
Immunoglobulin E (IgE) serum concentration regulator gene	IGES	I
Immunoglobulin epsilon (IgE)	IGHE	I
Immunoglobulin gamma (IgG) 2	IGHG2	I
Immunoglobulin heavy mu chain	IGHM	I
Immunoglobulin J polypeptide	IGJ	I
Immunoglobulin kappa constant region	IGKC	I
Immunoglobulin kappa variable region	IGKV	I
Insulin-like growth factor 1	IGF1	G
Insulin-like growth factor 1 receptor	IGF1R	G
Insulin-like growth factor 2	IGF2	G
Insulin-like growth factor 2 receptor	IGF2R	G
Integrin beta 1	ITGB1	G
Integrin beta 2	ITGB2	G
Integrin beta 3	ITGB3	G
Integrin beta 4	ITGB4	G
Integrin beta 5	ITGB5	G

Integrin beta 6	ITGB6	G
Integrin beta 7	ITGB7	G
Integrin, alpha 1	ITGA1	G
Integrin, alpha 2	ITGA2	G
Integrin, alpha 4	ITGA4	G
Integrin, alpha 5	ITGA5	G
Integrin, alpha 6	ITGA6	G
Integrin, alpha M	ITGAM	G
Intercellular adhesion molecule 1	ICAM1	I
Intercellular adhesion molecule 2	ICAM2	I
Intercellular adhesion molecule 3	ICAM3	I
Interferon alpha	IFNA1	I
Interferon beta	IFNB	I
Interferon gamma	IFNG	I
Interferon gamma receptor 1	IFNGR1	I
Interferon gamma receptor 2	IFNGR2	I
Interferon regulatory factor 1	IRF1	I
Interferon regulatory factor 4	IRF4	I
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) 10	IL10	I
Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I
Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
Janus kinase 1	JAK1	G

Janus kinase 2	JAK2	G
Janus kinase 3	JAK3	G
Kallikrein 3	KAK3	I
Kell blood group precursor	XK, KEL	T
Kininogen, High molecular weight	KNG	I
Kynureninase		E
Lactotransferrin	LTF	T
Laminin 5, alpha 3	LAMA3	G
Laminin 5, beta 3	LAMB3	G
Laminin 5, gamma 2	LAMC2	G
Laminin M	LAMM	G
Laminin receptor 1	LAMR1	G
Latent transforming growth factor-beta binding protein 2	LTBP2	G
Lectin, mannose-binding 1	LMAN1	I
Lectin, mannose-binding 2	MBL2	I
Leptin	LEP	G
Leptin receptor	LEPR	G
Leukaemia inhibitory factor	LIF	G
Leukaemia inhibitory factor receptor	LIFR	G
Leukin		I
Leukocyte-specific transcript 1	LST-1	I
Leukotriene A4 hydrolase		I
Leukotriene A4 synthase	LTA4S	E
Leukotriene B4 receptor		I
Leukotriene B4 synthase	LTB4S	E
Leukotriene C4 receptor		I
Leukotriene C4 synthase	LTC4S	E
Leukotriene D4/E4 receptor		I
LIM homeobox protein 1	LHX1	G
LIM homeobox protein 2	LHX2	G
LIM homeobox protein 3	LHX3	G
LIM homeobox protein 4	LHX4	G
LIM-domain only protein 1	LMO1	G
LIM-domain only protein 2	LMO2	G
LIM-domain only protein 3	LMO3	G
LIM-domain only protein 4	LMO4	G
LIM-Kinase I (LINK-I)		I
Lipocortin 1	ANX4	I
Lipoprotein-associated coagulation factor	LACI	I
Lipoxygenase 12 (platelets)	LOG12	I
Lipoxygenase 5 (leukocytes)		I
Lymphoblastic leukemia derived sequence 1	LYL1	I
Lymphocyte-specific protein tyrosine kinase	LCK	I
Lymphoid enhancer-binding factor	LEF-1	G
lymphotoxin		I
Lysozyme	LYZ	I
Macrophage activating factor	MAF	I

Macrophage inflammatory protein-1	MIP1	I
Macrophage inflammatory protein-1 receptor		I
Macrophage inflammatory protein-2	MIP2	I
Macrophage inflammatory protein-2 receptor		I
MAD (mothers against decapentaplegic, Drosophila) homologue 3	MADH3	G
MAD (mothers against decapentaplegic, Drosophila) homologue 4	MADH4	G
Malignant proliferation, eosinophil gene	MPE	I
Mannose binding protein	MBP	I
Mannosidase, alpha B lysosomal	MANB	E
Marenostrin	MEFV	T
Matrix metalloproteinase 1	MMP1	E
Matrix metalloproteinase 10	MMP10	E
Matrix metalloproteinase 11	MMP11	E
Matrix metalloproteinase 12	MMP12	E
Matrix metalloproteinase 13	MMP13	E
Matrix metalloproteinase 14	MMP14	E
Matrix metalloproteinase 15	MMP15	E
Matrix metalloproteinase 16	MMP16	E
Matrix metalloproteinase 17	MMP17	E
Matrix metalloproteinase 18	MMP18	E
Matrix metalloproteinase 19	MMP19	E
Matrix metalloproteinase 2	MMP2	E
Matrix metalloproteinase 3	MMP3, STMY1	E
Matrix metalloproteinase 4	MMP4	E
Matrix metalloproteinase 5	MMP5	E
Matrix metalloproteinase 6	MMP6	E
Matrix metalloproteinase 7	MMP7	E
Matrix metalloproteinase 8	MMP8	E
Matrix metalloproteinase 9	MMP9	E
MHC Class I: A		I
MHC Class I: B		I
MHC Class I: C		I
MHC Class I: LMP-2, LMP-7		I
MHC Class I: Tap1	ABCR, TAP1	I
MHC Class II: DP	HLA-DPB1	I
MHC Class II: DQ		I
MHC Class II: DR		I
MHC Class II: Tap2	TAP2, PSF2	I
MHC Class II:Complementation group A	MHC2TA	I
MHC Class II:Complementation group B	rfxank	I
MHC Class II:Complementation group C	RFX5	I
MHC Class II:Complementation group D	RFXAP	I
Monocyte chemoattractant protein 1	MCP1	I
Mucin 18	MUC18	T
Mutated in colorectal cancers, MCC	MCC	G
MutL homolog 1	MLH1	G

MutS homolog 2	MSH2	G
MutS homolog 3	MSH3	G
Myeloid leukemia factor-1	MLF1	I
Myeloperoxidase	MPO	I
Myoglobin		T
Myosin 5A	MYO5A	S
N-acyl hydrolase		I
NADPH oxidase		I
NADPH-dependent cytochrome P450 reductase	POR	E
Natural resistance-associated macrophage protein 1	NRAMP1	I
NB6		I
Nerve growth factor	NGF	G
Nerve growth factor receptor	NGFR	G
Neurofibromin 1	NF1	G
Neurofibromin 2	NF2	G
Neurokinin A	NKNA	N
Neurokinin B	NKNB	N
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Neutral endopeptidase		E
Neutrophil cystolic factor 1	NCF1	I
Neutrophil cystolic factor 2	NCF2	I
Nitric oxide synthase 1, NOS1	NOS1	E
Nitric oxide synthase 2, NOS2	NOS2	E
Nitric oxide synthase 3, NOS3	NOS3	E
Norrie disease protein	NDP	G
Notch 3	NOTCH3	G
Notch ligand - jagged 1	JAG1, AGS	G
Nuclear factor I-kappa-B-like gene	IKBL	I
Nuclear factor kappa beta	NFKB	I
Nuclear factor of activated T cells (NFAT) complex, cytosolic	NFATC	G
Nuclear factor of activated T cells (NFAT) complex, preexisting component	NFATP	G
Nucleoside diphosphate kinase-A	NDPKA	E
Oncogene bcl2		G
Oncogene ELK1	ELK1	G
Oncogene ELK2	ELK2	G
Oncogene ERG (early reponse gene)		G
Oncogene GLI1	GLI	G
Oncogene GLI2	GLI2	G
Oncogene GLI3	GLI3	G
Oncogene spi1		G
Oncogene TEL	ETV6	G
Oncostatin M	OSM	G



Oncostatin M receptor	OSMR	G
Ornithine delta-aminotransferase	OAT	E
Osteonectin	ON	G
Osteopontin	OPN	G
Paired box homeotic gene 3	PAX3	G
Paired box homeotic gene 7	PAX7	G
Patched (Drosophila) homolog, PTCH	PTCH	G
Peanut-like 1	PNUTL1	I
Phagocytin		I
Phenylethanolamine N-methyltransferase, PNMT	PNMT	E
Phosphatidylinositol glycan, class A (paroxysmal nocturnal hemoglobinuria)	PIGA	G
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C alpha		I
Phospholipase C beta		I
Phospholipase C delta	PLCD1	I
Phospholipase C epsilon		I
Phospholipase C gamma	PLCG1	I
Phosphomannomutase-2	PMM2	T
Plakophilin 1	PKP1	T
Plasminogen	PLG	E
Plasminogen activator inhibitor 1	PAI1	E
Plasminogen activator inhibitor 2	PAI2	E
Plasminogen activator receptor, Urokinase	UPAR; PLAUR	S
Plasminogen activator, Tissue	PLAT; TPA	E
Plasminogen activator, Urokinase	UPA; PLAU	E
Platelet glycoprotein 1b, alpha	GP1BA	I
Platelet glycoprotein 1b, beta	GP1BB	I
Platelet glycoprotein 1b, gamma	GP1BG	I
Platelet glycoprotein IX	GP9	I
Platelet glycoprotein V	GP5	I
Platelet-activating factor acetylhydrolase 1B	PAFAH1B1 or LIS1	I
Platelet-activating factor acetylhydrolase 2	PAFAH2	I
Platelet-activating factor receptor	PAFR	I
Poliovirus receptor	PVR, PVS	I
Potassium channel, calcium-activated,	KCNN4	N
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium inwardly-rectifying channel J11	KCNJ11	N
Potassium voltage-gated channel A1	KCNA1	N
Potassium voltage-gated channel E1	KCNE1	N

Potassium voltage-gated channel Q1	KCNQ1	N
Potassium voltage-gated channel Q2	KCNQ2	N
Potassium voltage-gated channel Q3	KCNQ3	N
Prekallikrein		I
Preproenkephalin	PENK	N
Procollagen N-protease		E
Promyelocytic leukemia gene	PML	G
Proopiomelanocortin	POMC	N
Properdin P factor, complement	PFC, PFD	I
Prostacyclin synthase		I
Prostaglandin (PG) D synthase, hematopoietic	PGDS	E
Prostaglandin 15-OH dehydrogenase	HGPD; PGDH	I
Prostaglandin D - DP receptor		I
Prostaglandin E1 receptor		I
Prostaglandin E2 receptor		I
Prostaglandin E3 receptor		I
Prostaglandin F - FP receptor		I
Prostaglandin I2 receptor		T
Prostaglandin IP receptor		I
Prostaglandin isomerase		G
Prostaglandin-endoperoxidase synthase 2	PTGS2	G
Protease inhibitor 1		T
Protein C	PROC	I
Protein C inhibitor	PCI	I
Protein kinase A		E
Protein kinase C, alpha	PRKCA	E
Protein kinase C, gamma	PRKCG	E
Protein kinase DNA-activated	PRKDC	E
Protein kinase G		E
Protein phosphatase 1, regulatory (inhibitor) subunit 3	PPP1R3	E
Protein phosphatase 2, regulatory subunit A, beta isoform	PPP2R1B	E
Protein S	PROS1	I
Protein tyrosine phosphatase, non-receptor type 12	PTPN12	G
Proteinase 3		I
Prothrombin precursor	F2	I
Purine nucleoside phosphorylase	NP	E
Pyruvate decarboxylase	PDHA	E
Retinoblastoma 1	RB1	G
Retinol binding protein 4	RBP4	T
Rhesus blood group, CcEe antigens	RHCE	T
Rhesus blood group, D antigen	RHD	T
Rhesus blood group-associated glycoprotein	RHAG	T
Ribosomal protein S19	RPS19	E
RIGUI	RIGUI	G
S100 calcium-binding protein A1	S100A1	N

S100 calcium-binding protein A2	S100A2	N
S100 calcium-binding protein A3	S100A3	N
S100 calcium-binding protein A4	S100A4	N
S100 calcium-binding protein A5	S100A5	N
S100 calcium-binding protein A6	S100A6	N
S100 calcium-binding protein A7	S100A7	N
S100 calcium-binding protein A8	S100A8	N
S100 calcium-binding protein A9	S100A9	N
S100 calcium-binding protein B	S100B	N
S100 calcium-binding protein P	S100P	N
SAP (SLAM-associated protein)	SH2D1A	I
Selectin E	SELE	N
Selectin L	SELL	N
Selectin P	SELP	N
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Severe combined immunodeficiency, type A (Athabaskan)	SCIDA	I
Signal transducer and activator of transcription 1	STAT1	G
Signal transducer and activator of transcription 2	STAT2	G
Signal transducer and activator of transcription 3	STAT3	G
Signal transducer and activator of transcription 4	STAT4	G
Signal transducer and activator of transcription 5	STAT5	G
Signaling lymphocyte activation molecule	SLAM	I
Sine oculis homeobox, drosophila, homolog 1	SIX1	G
Sine oculis homeobox, drosophila, homolog 2	SIX2	G
Sjogren (Sjogren) syndrome antigen A1	SSA1	I
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage gated, type V, alpha	SCN5A	N

polypeptide		
Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Solute carrier family 19 (folate transporter), member 1	SLC19A1	T
Solute carrier family 20, member 1	SLC20A1	T
Solute carrier family 20, member 2	SLC20A2	T
Solute carrier family 5 (sodium/glucose transporter), member 1	SLC5A1	T
Solute carrier family 5 (sodium/glucose transporter), member 2	SLC5A2	T
Solute carrier family 5 (sodium/glucose transporter), member 5	SLC5A5	T
Solute carrier family 5, member 3	SLC5A3	T
Sorcin	SRI	T
Sperm protamine P1	PRM1	G
Sperm protamine P2	PRM2	G
Stem cell factor	SCF	G
Stromal derived factor 1	SDF1	G
Succinate dehydrogenase 1	SDH1	E
Succinate thiokinase		E
Superoxide dismutase 1	SOD1	E
Superoxide dismutase 3	SOD3	E
SYK-related tyrosine kinase	SRK	I
Talin	TLN	G
Talin, TLN		S
T-cell acute lymphocytic leukemia 1	TAL1	I
T-cell acute lymphocytic leukemia 2	TAL2	I
T-cell receptor, alpha	TCRA	I
T-cell receptor, delta	TCRD	I
Tenascin (cytotactin)		S
Tenascin XA	TNXA	S
Terminal deoxynucleotidyltransferase	TDT	I
Terminal deoxynucleotidyltransferase, TDT		E
Thrombin receptor	F2R	I
Thrombopoietin	THPO	G
Thrombospondin	THBS1	G
Thromboxane A synthase 1	TBXAS1	I
Thromboxane A2	TXA2	I
Thromboxane A2 receptor	TBXA2R	I
Thy-1 T-cell antigen	THY1	I
Thymic humoral factor		I
Thymopoietin	TMPO	G
Thymosin		I
TIE receptor tyrosine kinase	TIE-1	G
Tip-associated protein	TAP	I
Toll-like receptor 4	TLR4	I
Topoisomerase I		E

Topoisomerase II		E
Transcobalamin 2, TCN2	TCN2	T
Transcription factor 3	TCF3	G
Transcription factor binding to IGHM enhancer 3	TFE3	G
Transferrin	TF	G
Transferrin receptor	TFRC	G
Transforming growth factor, alpha	TGFA	G
Transforming growth factor, beta 2	TGFB2	G
Transforming growth factor, beta induced	TGFB1	G
Transforming growth factor, beta receptor 2	TGFR2	G
Tuberous sclerosis 1	TSC1	G
Tuberous sclerosis 2	TSC2	G
Tubulin		S
Tumor susceptibility gene 101	TSG101	G
Tumour necrosis factor (TNF) receptor associated factor 1	TRAF1	I
Tumour necrosis factor (TNF) receptor associated factor 2	TRAF2	I
Tumour necrosis factor (TNF) receptor associated factor 3	TRAF3	I
Tumour necrosis factor (TNF) receptor associated factor 4	TRAF4	I
Tumour necrosis factor (TNF) receptor associated factor 5	TRAF5	I
Tumour necrosis factor (TNF) receptor associated factor 6	TRAF6	I
Tumour necrosis factor alpha	TNFA	I
Tumour necrosis factor alpha receptor	TNFAR	I
Tumour necrosis factor beta	TNFB	I
Tumour necrosis factor beta receptor	TNFR	I
Tumour protein p53	TP53, P53	G
Tumour protein p63	TP63	G
Tumour protein p73	TP73	G
Tumour protein, translationally-controlled 1	TPT1	G
Tumour suppressor gene DRA	DRA	I
Ubiquitin		G
Ubiquitin activating enzyme, E1		E
Ubiquitin B	UBB	G
Ubiquitin C	UBC	G
Ubiquitin fusion degeneration 1-like	UFD1L	G
Ubiquitin protein ligase E3A	UBE3A	E
Undulin 1	COL14A1	S
Uridine monophosphate kinase	UMPK	I
Uridine monophosphate synthetase	UMPS	I
Uroporphyrinogen III synthase	UROS	E
Vimentin	VIM	I
v-myc avian myelocytomatosis viral oncogene	MYC	G

homolog		
Von Hippel-Lindau gene	VHL	G
Werner syndrome helicase	WRN	G
Wilms tumour gene 1	WT1	G
Wilms tumour gene 2	WT2	G
Wilms tumour gene 4	WT4	G
Winged helix nude	WHN	G
Wiskott-Aldrich syndrome protein	WASP, THC	I
Xanthine dehydrogenase	XDH	E
X-ray repair gene	XRCC9	G
Zinc finger protein 198	ZIC198	S
Zinc finger protein HRX	ALL1	I

351. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 350.

352. A set according to claim 350 or 351 in which a minority of said probes for listed genes are absent.

353. A set according to claim 350 or 351 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.

354. A set according to claim 350 or 351 in which a limited number of probes are replaced by probes for non-listed genes.

355. A set of probes for a core group of genes according to any of claims 350 to 354 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.

356. A set according to any of claims 350 to 355 consisting of probes for members of a sub-group of the core group.

357. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.

358. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.

359. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.

360. A set according to claim 357 or 358 in which said substrate is a semiconductor microchip.

361. A set according to any preceding claim for use in a biological assay for detection of said gene variants.

362. A set according to any preceding claim for use in the measurement of differential gene expression levels.
363. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
364. A medical device including a set according to any of claims 350 to 362 for use in an array for detection of differential gene expression levels.
365. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 350) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 350 and 352 to 362 and relating the probe hybridisation pattern to said variations.
366. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 351) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 351 to 362 and relating the probe interaction pattern to said variations.
367. Use of a set or device according to any of claims 350 to 362 for the prognosis and management of patients suffering from or at risk of experiencing the symptoms and consequences of injury, inflammation, infection, immunity and/or repair.
368. Use of a set or device according to any of claims 350 to 362 for predicting likely therapeutic response and adverse events following therapeutic intervention.
369. Use of a set or device according to any of claims 350 to 362 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
370. Use of a set or device according to any of claims 350 to 362 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
371. Use of a set or device according to any of claims 350 to 362 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
372. Use of a set or device according to any of claims 350 to 362 for the development of new strategies of therapeutic intervention and in clinical trials.
373. Use of a set or device according to any of claims 350 to 362 for construction of and generation of algorithms for patient and healthcare management.
374. Use of a set or device according to any of claims 350 to 362 for modelling or assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations
375. Use of a set or device according to any of claims 350 to 362 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
376. Use of a set or device according to any of claims 350 to 362 for predicting optimum configuration/management of thereapeutic intervention.
377. A method according to claim 365 or 366 in which the identification of gene variants is indicative of a higher risk of experiencing the symptoms and

consequences of injury, inflammation, infection, immunity and/or repair for the patient or individual.

378. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop the symptoms and consequences of injury, inflammation, infection, immunity and/or repair which method comprises:
- i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from the symptoms and consequences of injury, inflammation, infection, immunity and/or repair;
  - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the symptoms and consequences of injury, inflammation, infection, immunity and/or repair;
  - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 350 to 356;
  - iv) calculating the frequencies of these alleles in the samples from i) and ii);
  - v) comparing the frequencies of these alleles in i) and ii);
  - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of experiencing the symptoms and consequences of injury, inflammation, infection, immunity and/or repair.
379. A method for assessing whether a given subject will be at risk of developing symptoms, which comprises comparing said subject's genotype with a model generated by the method of claim 278.
380. A method according to any of claims 365, 366, 378 and 379 wherein at least one step is computer-controlled.
381. An assay suitable for use in a method according to any of claims 365, 366, 378 and 379; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 350 to 356 in a biological sample.
382. A formatted assay technique (kit) for use in assessing the risk of a patient or individual experiencing the symptoms and consequences of injury, inflammation, infection, immunity and/or repair; said kit comprising:
- i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 350 or 352 to 356 in a sample of human DNA;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual experiencing the symptoms and consequences of injury, inflammation, infection, immunity and/or repair.
383. A formatted assay technique (kit) for use in assessing the risk of a patient or individual experiencing the symptoms and consequences of injury, inflammation, infection, immunity and/or repair; said kit comprising:
- i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core group of genes as defined in any of claims 351 to 356 in an expressed-protein-containing human sample;
  - ii) reagents for use in the detection process



- iii) readout indicating the probability of a patient or individual experiencing the symptoms and consequences of injury, inflammation, infection, immunity and/or repair.

384. A set of probes according to claim 350, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.

385. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to dysfunction, damage or disease consequent on an aberration in the processes of development; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

#### KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME  
T TRANSPORT & STORAGE  
S STRUCTURAL  
I IMMUNITY  
N NERVOUS TRANSMISSION  
G GROWTH & DIFFERENTIATION

#### DEVELOPMENT GENE LIST

	HUGO gene symbol	Protein function
17-ketosteroid reductase		N
2,4-dienoyl CoA reductase	DECR	E
3 beta hydroxysteroid dehydrogenase 2	HSD3B2	E
3-oxoacid CoA transferase	OXCT	E
6-pyruvoyltetrahydropterin synthase	PTS	E
Absent in melanoma 1 gene	AIM1	G
Acetoacetyl 2-CoA-thiolase	ACAT2	E
Acetyl CoA acyltransferase	ACAA	E
Acetyl CoA carboxylase alpha	ACACA	E
Acetylcholine receptor, nicotinic, alpha A1	CHRNA1	N
Acetylcholine receptor, nicotinic, alpha A2	CHRNA2	N
Acetylcholine receptor, nicotinic, alpha A3	CHRNA3	N
Acetylcholine receptor, nicotinic, alpha A4	CHRNA4	N
Acetylcholine receptor, nicotinic, alpha A5	CHRNA5	N
Acetylcholine receptor, nicotinic, alpha A6	CHRNA6	N
Acetylcholine receptor, nicotinic, alpha A7	CHRNA7	N
Acetylcholine receptor, nicotinic, beta 1	CHRNA1	N
Acetylcholine receptor, nicotinic, beta 2	CHRNA2	N
Acetylcholine receptor, nicotinic, beta 3	CHRNA3	N
Acetylcholine receptor, nicotinic, beta 4	CHRNA4	N

Acetylcholine receptor, nicotinic, epsilon	CHRNE	N
Acetylcholine receptor, nicotinic, gamma	CHRNA3	N
Acetylcholinesterase	ACHE	E
Achromatopsia 2	ACHM2	S
Acid phosphatase 2, lysosomal	ACP2	E
Acrosin	ACR	G
Actin, alpha, cardiac	ACTC	S
Actin, alpha, skeletal	ACTA1	S
Actin, alpha, smooth, aortic	ACTA2	S
Activin		G
Activin A receptor, type 2B	ACVR2B	G
Activin A receptor, type 2-like kinase 1	ACVRL1	G
Acyl CoA dehydrogenase, short chain	ACADS	E
Acyl-CoA thioesterase		E
ADAM (A disintegrin and metalloproteinase) 1	ADAM1	E
ADAM (A disintegrin and metalloproteinase) 10	ADAM10	E
ADAM (A disintegrin and metalloproteinase) 11	ADAM11	E
ADAM (A disintegrin and metalloproteinase) 12	ADAM12	E
ADAM (A disintegrin and metalloproteinase) 13	ADAM13	E
ADAM (A disintegrin and metalloproteinase) 14	ADAM14	E
ADAM (A disintegrin and metalloproteinase) 15	ADAM15	E
ADAM (A disintegrin and metalloproteinase) 16	ADAM16	E
ADAM (A disintegrin and metalloproteinase) 17	ADAM17	E
ADAM (A disintegrin and metalloproteinase) 18	ADAM18	E
ADAM (A disintegrin and metalloproteinase) 19	ADAM19	E
ADAM (A disintegrin and metalloproteinase) 2	ADAM2	E
ADAM (A disintegrin and metalloproteinase)	ADAM3A	E
ADAM (A disintegrin and metalloproteinase)	ADAM3B	E
ADAM (A disintegrin and metalloproteinase) 4	ADAM4	E
ADAM (A disintegrin and metalloproteinase) 5	ADAM5	E
ADAM (A disintegrin and metalloproteinase) 6	ADAM6	E
ADAM (A disintegrin and metalloproteinase) 7	ADAM7	E
ADAM (A disintegrin and metalloproteinase) 8	ADAM8	E
ADAM (A disintegrin and metalloproteinase) 9	ADAM9	E
Adducin, alpha	ADD1	S
Adducin, beta	ADD2	S
Adenomatous polyposis coli tumour suppressor gene	APC	G
Adenosine deaminase	ADA	E
Adenosine monophosphate deaminase	AMPD	E
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N
Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenyl cyclase		N
Adenylate cyclase 1	ADCY1	E

Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adenylosuccinate lyase	ADSL	E
ADP-ribosyltransferase	ADPRT	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
Adrenocorticotrophic hormone (ACTH) receptor	ACTHR	G
Adrenoleukodystrophy gene	ALD	E
Alanine-glyoxylate aminotransferase	AGXT	E
Albumin, ALB	ALB	T
Aldehyde dehydrogenase 1	ALDH1	E
Aldehyde dehydrogenase 10	ALDH10	E
Aldehyde dehydrogenase 2	ALDH2	E
Aldehyde dehydrogenase 5	ALDH5	E
Aldehyde dehydrogenase 6	ALDH6	E
Aldehyde dehydrogenase 7	ALDH7	E
Aldolase A	ALDOA	E
Aldolase B	ALDOB	E
Aldolase C	ALDOC	E
Aldosterone receptor	MLR	G
Alkaline phosphatase, liver/bone/kidney	ALPL	T
Alkaptonuria gene	AKU	G
Alkylglycerone phosphate synthase	AGPS	E
Alpha 2 macroglobulin	A2M	I
alpha tectorin	TECTA	G
alpha thalassemia gene	ATR $\times$	N
alpha1-antitrypsin	PI	E
alpha2-antiplasmin	PLI	E
alpha-actinin 2	ACTN2	G
alpha-actinin 3	ACTN3	G
alpha-amylase		E
Alpha-fetoprotein	AFP	G
alpha-Galactosidase A	GLA	E
alpha-ketoglutarate dehydrogenase		E
alpha-L-Iduronidase	IDUA	E
alpha-synuclein	SNCA	N
Amelogenin	AMELX	S
Aminomethyltransferase	AMT	E

Aminopeptidase P	XPNPEP2	E
Amphiregulin	AREG	G
Amylo-1,6-glucosidase	AGL	E
Amyloid beta (A4) precursor protein-binding, APBB1	APBB1	N
Amyloid beta A4 precursor protein	APP	N
Amyloid beta A4 precursor-like protein	APLP	N
Androgen binding protein	ABP	T
Androgen receptor	AR	G
Angiopoietin 1	ANGPT1	G
Angiopoietin 2	ANGPT2	G
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensinogen	AGT	E
Ankyrin 1	ANK1	S
Ankyrin 2	ANK2	S
Ankyrin 3	ANK3	S
Antidiuretic hormone receptor	ADHR	T
Anti-Mullerian hormone	AMH	G
Anti-Mullerian hormone type 2 receptor	AMHR2	G
Antithrombin III	AT3	E
AP-2, alpha	TFAP2A	G
AP-2, beta	TFAP2B	G
AP-2, gamma	TFAP2C	G
Apaf-1		S
Apical protein, xenopus laevis-like	APXL	G
Apolipoprotein A 4	APOA4	T
Apolipoprotein A I	APOA1	T
Apolipoprotein A II	APOA2	T
Apolipoprotein B	APOB	T
Apolipoprotein C1	APOC1	T
Apolipoprotein C2	APOC2	T
Apolipoprotein C3	APOC3	T
Apolipoprotein D	APOD	T
Apolipoprotein E	APOE	T
Apolipoprotein H	APOH	T
Apopain	CPP32	G
Apoptosis antigen 1	APT1	I
Apoptosis antigen ligand 1	APT1LG1	I
Apoptosis-inducing factor	AIF	I
Apurinic endonuclease	APE	E
Archaete-scute homolog 1	ASH1	G
Archaete-scute homolog 2	ASH2	G
Arginosuccinate synthetase	ASS	E
Arrestin	SAG	S
Aryl hydrocarbon receptor	AHR	T
Aryl hydrocarbon receptor nuclear translocator	ARNT	T
Arylsulfatase A	ARSA	E
Arylsulfatase B	ARSB	E

Arylsulfatase C	ARSC1	E
Arylsulfatase D	ARSD	E
Arylsulfatase E	ARSE	E
Arylsulfatase F	ARSF	E
Aspartate transaminase		T
Aspartate transcarbamoylase		E
Aspartoacylase	ASPA	E
Aspartylglucosaminidase	AGA	E
Astrotactin	ASTN	G
Ataxia telangiectasia complementation group D	ATD, ATDC	G
Ataxia telangiectasia gene, AT	ATM	G
Ataxin 1	SCA1	G
Ataxin 2	SCA2	G
Ataxin 3	MJD	G
ATP-binding cassette transporter 7	ABC7	I
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G
Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
Atrophin 1	DRPLA	G
Attractin		I
Autoimmune regulator, AIRE	AIRE	I
Azoospermia factor 1	AZF1	G
Bagpipe homeobox, drosophila homolog of, 1	BAPX1	G
B-cell CLL/lymphoma 1	BCL1	I
B-cell CLL/lymphoma 10	BCL10	I
B-cell CLL/lymphoma 3	BCL3	I
B-cell CLL/lymphoma 4	BCL4	I
B-cell CLL/lymphoma 5	BCL5	I
B-cell CLL/lymphoma 6	BCL6	I
B-cell CLL/lymphoma 7	BCL7	I
B-cell CLL/lymphoma 8	BCL8	I
B-cell CLL/lymphoma 9	BCL9	I
BCL2-associated X protein	BAX	G
BCL2-related protein A1	BCL2A1	G
Beckwith-Wiedemann region 1A	BWR1A	G
Bestrophin	VMD2	T
beta 2 microglobulin	B2M	I
beta-endorphin receptor		N
beta-Glucuronidase	GUSB	E
beta-N-acetylhexosaminidase, A		E
beta-N-acetylhexosaminidase, B		E
Bilirubin UDP-glucuronosyltransferase		E
Bleomycin hydrolase	BLMH	E
Bloom syndrome protein	BLM	G
Blue cone pigment	BCP	S
Bone morphogenetic protein, BMP1	BMP1	G
Bone morphogenetic protein, BMP2	BMP2	G

Bone morphogenetic protein, BMP3	BMP3	G
Bone morphogenetic protein, BMP4	BMP4	G
Bone morphogenetic protein, BMP5	BMP5	G
Bone morphogenetic protein, BMP6	BMP6	G
Bone morphogenetic protein, BMP7	BMP7	G
Bone morphogenetic protein, BMP8	BMP8	G
Brain derived neurotrophic factor	BDNF	G
Brain derived neurotrophic factor (BDNF) receptor	BDNFR	G
Branched chain aminotransferase 1, cytosolic	BCAT1	E
Branched chain aminotransferase 2, mitochondrial	BCAT2	E
BRCA1-associated RING domain gene 1	BARD1	G
Breakpoint cluster region	BCR	G
Breast cancer 1	BRCA1	G
Breast cancer 2	BRCA2	G
Breast cancer, ductal, 1	BRCD1	G
Breast cancer, ductal, 2	BRCD2	G
Bruton agammaglobulinaemia tyrosine kinase	BTK	G
Butyrylcholinesterase	BCHE	E
C3 convertase		E
Ca(2+) transporting ATPase, fast twitch	ATP2A1	T
Ca(2+) transporting ATPase, slow twitch	ATP2A2	T
Cadherin E	CDH1	G
Cadherin EP		G
Cadherin N	CDH2	G
Cadherin P	CDH3	G
Calbindin 1	CALB1	G
Calbindin D9K	CALB3	G
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha-1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha-1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha-1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha-1E (CACNL1A6)	CACNA1E	N
Calcium channel, voltage-dependent, Alpha-2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, L type, alpha 1S subunit	CACNA1S	N
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N
Calcium channel, voltage-dependent, P/Q	CACNA1A	N

type, alpha 1A subunit		
Calcium channel, voltage-dependent, T-type		N
Calcium sensing receptor	CASR	T
Calmodulin 1	CALM1	G
Calmodulin 2	CALM2	G
Calmodulin 3	CALM3	G
Calmodulin dependant kinase		T
Calmodulin-dependant protein kinase II	CAMK2A	G
Calnexin	CANX	G
Calpain	CAPN, CAPN3	E
Canalicular multispecific organic anion transporter	CMOAT	T
Carbamoylphosphate synthetase 1	CPS1	E
Carbamoylphosphate synthetase 2	CPS2	E
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Cardiac-specific homeobox, CSX	CSX	G
Carnitine acetyltransferase	CRAT	E
Carnitine acylcarnitine translocase	CACT	E
Carnitine transporter protein	CDSP, SCD	T
Cartilage oligomeric matrix protein	COMP, EDM1, PSACH	N
Cartilage-hair hypoplasia gene	CHH	N
Caspase 1	CASP1	G
Caspase 10	CASP10	G
Caspase 2	CASP2	G
Caspase 3	CASP3	G
Caspase 4	CASP4	G
Caspase 5	CASP5	G
Caspase 6	CASP6	G
Caspase 7	CASP7	G
Caspase 8	CASP8	G
Caspase 9	CASP9	G
Catechol-O-methyltransferase	COMT	E
Catenin, alpha	CTNNA1	G
Catenin, beta	CTNNB1	G
Catenin, gamma		G
Cathepsin K	CTSK	E
Caveolin 3	CAV3	E
CD1	CD1	I
CD44	CD44	I
Cdc 25 phosphatase		G
Cdc2	CDC2	G
CDX1		G
CEA		G
Cell adhesion molecule, intercellular, ICAM	ICAM1	G

Cell adhesion molecule, leukocyte-endothelial, LECAM (CD62)	LECAM1	G
Cell adhesion molecule, liver, LCAM	LCAM	G
Cell adhesion molecule, neural, NCAM1	NCAM1	G
Cell adhesion molecule, neural, NCAM120	NCAM120	G
Cell adhesion molecule, neural, NCAM2	NCAM2	G
Cell adhesion molecule, platelet-endothelial, PECAM	PECAM1	G
Cell adhesion molecule, vascular, VCAM	VCAM1	G
Cellubrevin	CEB	N
c-erbB1	ERBB1	G
c-erbB2	ERBB2	G
c-erbB3	ERBB3	G
c-erbB4	ERBB4	G
Ceroid lipofuscinosis neuronal 2	CLN2	N
Ceroid lipofuscinosis neuronal 3	CLN3	N
Ceroid lipofuscinosis neuronal 4	CLN4	N
Ceroid lipofuscinosis neuronal 5	CLN5	N
Ceroid lipofuscinosis neuronal 6	CLN6	N
Chediak-Higashi syndrome 1 gene	CHS1	T
Chemokine MCAF	MCAF	I
Chemokine receptor CCR2	CCR2	I
Chemokine receptor CCR3	CCR3	I
Chemokine receptor CCR5	CCR5	I
Chemokine receptor CXCR1	CXCR1	I
Chemokine receptor CXCR2	CXCR2	I
Chemokine receptor CXCR4	CXCR4	I
Chloride channel 5	CLCN5	S
Cholestasis, progressive familial intrahepatic 1 gene	FIC1	G
Cholesterol ester transfer protein	CETP	T
Choline acetyltransferase	CHAT	E
Choroideremia gene	CHM	S
Chromogranin A	CHGA	G
Ciliary neurotrophic factor (CNTF)	CNTF	G
Ciliary neurotrophic factor (CNTF) receptor	CNTFR	G
c-kit receptor tyrosine kinase		G
Clathrin		T
Cleavage signal-1 protein	CS1	G
Cleft palate gene	CPX	G
Clusterin	CLU	G
CoA transferase		E
Cochlin	COCH	I
Cockayne syndrome gene, CKN1	CKN1	G
Collagen I alpha 1	COL1A1	S
Collagen I alpha 2	COL1A2	S
Collagen II alpha 1	COL2A1	S
Collagen III alpha 1	COL3A1	S



Collagen IV alpha 1	COL4A1	S
Collagen IV alpha 2	COL4A2	S
Collagen IV alpha 3	COL4A3	S
Collagen IV alpha 4	COL4A4	S
Collagen IV alpha 5	COL4A5	S
Collagen IV alpha 6	COL4A6	S
Collagen IX alpha 2	COL9A2, EDM2	S
Collagen IX alpha 3	COL9A3	S
Collagen receptor	COLR	S
Collagen V alpha 1	COL5A1	S
Collagen V alpha 2	COL5A2	S
Collagen VI alpha 1	COL6A1	S
Collagen VI alpha 2	COL6A2	S
Collagen VI alpha 3	COL6A3	S
Collagen VII alpha 1	COL7A1	S
Collagen X alpha 1	COL10A1	S
Collagen X alpha 1	COL11A1	S
Collagen XI alpha 2	COL11A2	S
Collagen XVII alpha 1	COL17A1	S
Collagenic-like tail subunit of asymmetric acetylcholinesterase	COLQ	E
Collapsin		G
Colony-stimulating factor 1	CSF1	G
Colony-stimulating factor 1 receptor	CSF1R	G
Colony-stimulating factor 2	CSF2	G
Colony-stimulating factor 2 alpha receptor	CSF2RA	G
Colony-stimulating factor 2 beta receptor	CSF2RB	G
Colony-stimulating factor 3	CSF3	G
Colony-stimulating factor 3 receptor	CSF3R	G
Complex V	MTATP6	E
Cone-rod homeobox-containing gene	CRX	G
Contactin	CNTN1	G
Core-binding factor, alpha 1	CBFA1	G
Core-binding factor, alpha 2	CBFA2	G
Core-binding factor, beta	CBFB	G
Corticotrophin-releasing hormone	CRH	T
Corticotrophin-releasing hormone receptor	CRHR1	T
Creatine kinase – B and m	CKBE	E
Creb binding protein	CREBBP	G
Cryptochrome 1	CRY1	S
Cryptochrome 2	CRY2	S
Crystallin, alpha A	CRYAA	S
Crystallin, alpha B	CRYAB	S
Crystallin, beta B2	CRYBB2	S
Crystallin, gamma A	CRYGA	S
c-src tyrosine kinase	CSK	G
Cu <sup>2+</sup> transporting ATPase alpha polypeptide	ATP7A	E
Cu <sup>2+</sup> transporting ATPase beta polypeptide	ATP7B	E

Cubilin	CUBN	T
Cyclic AMP response element binding protein	CREB	G
Cyclic AMP response element modulator	CREM	G
Cyclic AMP-dependent protein kinase	PKA	E
Cyclic nucleotide gated channel alpha 1, CNGA1	CNGA1	N
Cyclic nucleotide gated channel alpha 3, CNGA3	CNGA3	N
Cyclic nucleotide phosphodiesterase 1B	PDE1B	E
Cyclic nucleotide phosphodiesterase 1B1	PDE1B1	E
Cyclic nucleotide phosphodiesterase 2A3	PDE2A3	E
Cyclic nucleotide phosphodiesterase 3A	PDE3A	E
Cyclic nucleotide phosphodiesterase 3B	PDE3B	E
Cyclic nucleotide phosphodiesterase 4A	PDE4A	E
Cyclic nucleotide phosphodiesterase 4C	PDE4C	E
Cyclic nucleotide phosphodiesterase 5A	PDE5A	E
Cyclic nucleotide phosphodiesterase 6A	PDE6A	E
Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E
Cyclic nucleotide phosphodiesterase 8	PDE8	E
Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclin A	CCNA	G
Cyclin B	CCNB	G
Cyclin C	CCNC	G
Cyclin D	CCND1	G
Cyclin E	CCNE	G
Cyclin F	CCNF	G
Cyclin-dependent kinase 1	CDK1	G
Cyclin-dependent kinase 10	CDK10	G
Cyclin-dependent kinase 2	CDK2	G
Cyclin-dependent kinase 3	CDK3	G
Cyclin-dependent kinase 4	CDK4	G
Cyclin-dependent kinase 5	CDK5	G
Cyclin-dependent kinase 6	CDK6	G
Cyclin-dependent kinase 7	CDK7	G
Cyclin-dependent kinase 8	CDK8	G
Cyclin-dependent kinase 9	CDK9	G
Cyclin-dependent kinase inhibitor 1A (P21, CIP1)	CDKN1A	G
Cyclin-dependent kinase inhibitor 1B (P27, KIP1)	CDKN1B	G
Cyclin-dependent kinase inhibitor 1C (P57, KIP2)	CDKN1C	G
Cyclin-dependent kinase inhibitor 2A (p16)	CDKN2A	G
Cyclin-dependent kinase inhibitor 3	CDKN3	G
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
CYP11A1	CYP11A1	E

CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E
CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E
CYP2C18	CYP2C18	E
CYP2C19	CYP2C19	E
CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E
CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E
CYP2F1	CYP2F1	E
CYP2J2	CYP2J2	E
CYP3A3	CYP3A3	E
CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystathionase	CTH	E
Cystathione beta synthase	CBS	E
Cystic fibrosis transmembrane conductance regulator, CFTR	CFTR	N
Cystinosin	CTNS	T
Cytidine deaminase	CDA	E
Cytochrome b-245 alpha	CYBA	E
Cytochrome b-245 beta	CYBB	E
Cytochrome b-5	CYB5	E
DAX1 nuclear receptor	DAX1	I
Deafness autosomal dominant 5	DFNA5	N

Deafness dystonia peptide	DDP	N
Defender against cell death 1	DAD1	G
Deleted in azoospermia	DAZ	G
Deleted in colorectal carcinoma	DCC	G
Deleted in malignant brain tumours 1	DMBT1	G
Delta aminolevulinate dehydratase	ALAD	E
Delta(4)-3-oxosteroid 5-beta-reductase		E
Delta-7-dehydrocholesterol reductase	DHCR7	E
Dentin sialophosphoprotein	DSPP	G
Deoxyuridine triphosphatase; dUTPase		E
Desert hedgehog, dhh		G
DHEA sulfotransferase	STD	E
Diaphanous 1	DIAPH1	N
Diaphanous 2	DIAPH2	N
Diastrophic dysplasia sulfate transporter	DTD	T
Dihydrolipoamide branched chain transacylase	DBT	N
Dihydrolipoamide dehydrogenase	DLD	N
Dihydrolipoyl dehydrogenase 2	PDHA	E
Dihydrolipoyl transacetylase	PDHA	E
Dihydroorotase		E
Dihydroxyacetonephosphate acyltransferase	DHAPAT	E
Disrupted meiotic cDNA 1, homolog	DMC1	G
Distal-less homeobox 1	DLX1	G
Distal-less homeobox 2	DLX2	G
Distal-less homeobox 3	DLX3	G
Distal-less homeobox 4	DLX4	G
Distal-less homeobox 5	DLX5	G
Distal-less homeobox 6	DLX6	G
DNA damage binding protein, DDB1	DDB1	S
DNA damage binding protein, DDB2	DDB2	S
DNA directed polymerase, alpha	POLA	E
DNA glycosylases		E
DNA helicases		E
DNA Ligase 1	LIG1	E
DNA methyltransferase	DNMT	E
DNA polymerase 1		E
DNA polymerase 2		E
DNA polymerase 3		E
DNA primase		E
DNA-damage-inducible transcript 3	DDIT3	S
DNA-dependant RNA polymerase		E
DOPA decarboxylase	DDC	E
Doublecortin, DCX	DCX	S
Duffy blood group	FY	T
Dynamin	DNM1	G
Dynein		G
Dyskerin	DKC1	S
Dystonia 1	DYT1	S

Dystonia 3	DYT3	S
Dystonia 6	DYT6	S
Dystonia 7	DYT7	S
Dystonia 9	CSE	S
Dystrophia myotonica	DM, DMPK	E
Dystrophia myotonica, atypical	DM2	E
Dystrophin	DMD	S
Dystrophin-associated glycoprotein 35kD, SCGD	SGCD	S
Dystrophin-associated glycoprotein 35kD, SGSG	SGCG	S
Dystrophin-associated glycoprotein 43kD	SGCB	S
Dystrophin-associated glycoprotein 50kD	SGCA	S
E74-like factor 1, ELF1	ELF1	G
EB1		G
Ectodermal Dysplasia 1 gene	ED1	S
Electron-transferring-flavoprotein alpha	ETFA	T
Electron-transferring-flavoprotein beta	ETFB	T
Electron-transferring flavoprotein dehydrogenase	ETFDH	E
Empty spiracles (drosophila) homologue 1	EMX1	G
Empty spiracles (drosophila) homologue 2	EMX2	G
Endobrevin	VAMP8	N
Endocardial fibroelastosis 2 gene	EFE2	S
Endometrial bleeding-associated factor	EBAF	G
Endothelin 1	EDN1	N
Endothelin 2	EDN2	N
Endothelin 3	EDN3	N
Endothelin converting enzyme	ECE1	N
Endothelin receptor type A	EDNRA	N
Endothelin receptor type B	EDNRB	N
Engrailed-1	EN1	G
Engrailed-2	EN2	G
Enolase	ENO1	E
Enoyl CoA isomerase		E
Enterokinase	PRSS7, ENTK	E
Ephrin receptor tyrosine kinase A	EPHA	G
Ephrin receptor tyrosine kinase B	EPHB	G
Ephrin-A	EFNA	G
Ephrin-B	EFNB	G
Epidermal growth factor	EGF	G
Epidermal growth factor receptor	EGFR	G
Epilepsy, benign neonatal 4 gene	ICCA	E
Epilepsy, female restricted	EFMR	E
Epilepsy, progressive myoclonic 2 gene	EPM2A	E
Erythrocyte membrane protein band 4.1	EPB41	S
Erythrocyte membrane protein band 4.2	EPB42	S
Erythrocyte membrane protein band 7.2	EPB72	S

Erythroid kruppel-like factor	EKLF	G
Erythropoietin	EPO	I
Erythropoietin receptor	EPOR	I
Estrogen receptor	ESR	G
Eukaryotic initiation translation factor	EIF4E	G
EWS RNA-binding protein	EWSR1	G
Excision repair complementation group 1 protein	ERCC1	E
Excision repair complementation group 2 protein	ERCC2	E
Excision repair complementation group 2 protein	ERCC3	E
Excision repair complementation group 4 protein	ERCC4	E
Excision repair complementation group 6 protein	ERCC6	E
Exostosin 1	EXT1	S
Exostosin 2	EXT2	S
Exostosin 3	EXT3	S
Eyes absent 1	EYA1	G
Eyes absent 2	EYA2	G
Eyes absent 3	EYA3	G
Faciogenital dysplasia	FGD1, FGDY	T
Factor 1 (No. one)	F1	I
Factor B, properdin		I
Factor D		I
Factor H	HF1	I
Factor I (letter I)	IF	I
Factor III	F3	I
Factor IX	F9	I
Factor V	F5	I
Factor VII	F7	I
Factor VIII	F8	I
Factor X	F10	I
Factor XI	F11	I
Factor XII	F12	I
Factor XIII A & B	F13A & F13B	I
Fanconi anemia, complementation group A	FANCA	T
Fanconi anemia, complementation group C	FANCC	T
Fanconi anemia, complementation group D	FANCD	T
Fc fragment of IgG, high affinity IA, receptor for FCGR1A		G
Fc fragment of IgG, low affinity IIa, receptor for FCGR2A (CD32)		G
Fc fragment of IgG, low affinity IIIa, receptor for FCGR3A (CD16)		G
Fc receptor		I
Fertilin protein	FTNB	G
Fibrillin 1	FBN1	G

Fibrillin 2	FBN2	G
Fibroblast growth factor	FGF1	G
Fibroblast growth factor receptor 1	FGFR1	G
Fibroblast growth factor receptor 2	FGFR2	G
Fibroblast growth factor receptor 3	FGFR3	G
Fibronectin precursor	FN1	G
Flavin-containing monooxygenase 1	FMO1	E
Flavin-containing monooxygenase 2	FMO2	E
Flavin-containing monooxygenase 3	FMO3	E
Flavin-containing monooxygenase 4	FMO4	E
Flightless-II, Drosophila homolog of	FLII	G
Folic acid receptor	FOLR	G
Follicle stimulating hormone receptor	FSHR, ODG1	G
Follicle stimulating hormone, FSH	FSHB	G
Follicular lymphoma variant translocation 1	FVT1	I
Follistatin		G
Forkhead rhabdomyosarcoma gene	FKHR	G
Forkhead transcription factor 10	FKHL10	G
Forkhead transcription factor 14	FKHL14	G
Forkhead transcription factor 7	FKHL7	G
Formiminotransferase		E
Fragile site, folic acid type, rare, fra(X) A	FRAXA	N
Fragile site, folic acid type, rare, fra(X) E	FRAXE	N
Fragile site, folic acid type, rare, fra(X) F	FRAXF	N
Frataxin	FRDA	G
Fringe secreted protein, lunatic	LFNG	G
Fringe secreted protein, manic	MFNG	G
Fringe secreted protein, radical	RFNG	G
Fructose-1,6-diphosphatase	FBP1	E
Fucosyltransferase 6	FUT6	T
Fukuyama type congenital muscular dystrophy	FCMD	G
Fumarase	FH	E
Fumarylacetoacetase	FAH	E
G/T mismatch binding protein	GTBP, MSH6	G
GABA receptor, alpha 1	GABRA1	N
GABA receptor, alpha 2	GABRA2	N
GABA receptor, alpha 3	GABRA3	N
GABA receptor, alpha 4	GABRA4	N
GABA receptor, alpha 5	GABRA5	N
GABA receptor, alpha 6	GABRA6	N
GABA receptor, beta 1	GABRB1	N
GABA receptor, beta 2	GABRB2	N
GABA receptor, beta 3	GABRB3	N
GABA receptor, gamma 1	GABRG1	N
GABA receptor, gamma 2	GABRG2	N
GABA receptor, gamma 3	GABRG3	N
GABA transaminase	ABAT	E
Gadd45 (growth arrest & DNA-damage-inducible protein)		E

Galactocerebrosidase	GALC	E
Galactokinase	GALK1	E
Galactose 1-phosphate uridyl-transferase	GALT	E
Galactosyltransferase 1	GT1	G
Galactosyltransferase, alpha 1,3	GGTA1	G
Galactosyltransferase, beta 3	B3GALT	G
Galanin	GAL	N
Galanin receptor	GALNR1	N
Gamma-glutamyl carboxylase	GGCX	T
Gap junction protein alpha 1	GJA1	T
Gap junction protein alpha 3	GJA3	T
Gap junction protein alpha 8	GJA8	T
Gap junction protein beta 1	GJB1	T
Gap junction protein beta 2	GJB2	T
Gap junction protein beta 3	GJB3	T
Gastric Intrinsic factor, GIF	GIF	E
Gastrin	GAS	G
Gastrin releasing peptide	GRP	T
Gastrointestinal tumor-associated antigen 1	GA733	I
Gastrulation brain homeobox 2	GBX2	G
GDP dissociation inhibitor 1	GDI1	G
Gelsolin	GSN	G
Geniospasm 1	GSM1	G
Gephyrin		N
Glial-cell derived neurotrophic factor (GDNF) receptor		N
Glial-cell derived neurotrophic factor, GDNF	GDNF	N
Glioma chloride ion channel, GCC		G
Glucagon receptor	GCGR	G
Glucagon-like peptide receptor 1	GLP1R	G
Glucocorticoid receptor	GRL	G
Glucose-6-phosphatase translocase	G6PT1	E
Glucosidase, acid alpha	GAA	E
Glucosidase, acid beta	GBA	E
Glutamate decarboxylase, GAD	GAD1	E
Glutamate-cysteine ligase	GLCLC	E
Glutathione	GSH	T
Glutathione peroxidase, GPX1	GPX1	E
Glutathione peroxidase, GPX2	GPX2	E
Glutathione reductase, GSR	GSR	E
Glutathione S-transferase mu 1, GSTM1	GSTM1	E
Glutathione S-transferase mu 4, GSTM4		E
Glutathione S-transferase theta 1, GSTT1	GSTT1	E
Glutathione S-transferase theta 2, GSTT2		E
Glutathione S-transferase, GSTP1	GSTP1	E
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glutathione synthetase	GSS	E
Glyceraldehyde-3-phosphate dehydrogenase,	GAPDH	E



GAPDH		
Glycerol kinase	GK	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
Glycine dehydrogenase	GLDC	E
Glycine receptor, alpha	GLRA2	N
Glycine receptor, beta		N
Glycogen branching enzyme	GBE1	E
Glycogen phosphorylase	PYGL	E
Glycogen synthase 1 (muscle)	GLYS1	E
Glycogen synthase 2 (liver)	GYS2	E
Glycosyltransferases, ABO blood group	ABO	E
Glypican 3	GPC3, SDYS	G
GM2 ganglioside activator protein, GM2A	GM2A	E
Gonadotropin releasing hormone	GNRH	G
Gonadotropin releasing hormone receptor	GNRHR	G
Goosecoid GSC		G
Green cone pigment	GCP	S
Growth arrest-specific homeobox	GAX	G
Growth factor receptor-bound protein 2	GRB2	G
Growth hormone 1	GH1	G
Growth hormone 2 (placental)	GH2	G
Growth hormone receptor	GHR	G
Growth hormone releasing hormone (GHRH)	GHRH	G
Growth hormone releasing hormone receptor	GHRHR	G
Growth/differentiation factor 5	GDF5	G
Growth-regulated protein precursor, GRO	GRO	I
GTP cylcohydrolase 1	GCH1	G
GTPase-activating protein, GAP	RASA1	G
Guanidinoacetate N-methyltransferase	GAMT	E
Guanine nucleotide-binding protein, alpha activating activity polypeptide, GNAO	GNAO1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 1, GNAI1	GNAI1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 2, GNAI2	GNAI2	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 3, GNAI3	GNAI3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS1	GNAS1	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS2	GNAS2	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS3	GNAS3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS4	GNAS4	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT1	GNAT1	N

Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT2	GNAT2	N
Guanine nucleotide-binding protein, beta polypeptide 3	GNB3	N
Guanine nucleotide-binding protein, gamma polypeptide 5	GNG5	N
Guanine nucleotide-binding protein, q polypeptide	GNAQ	N
Guanylate cyclase 2D, membrane (retina-specific)	GUCY2D	E
Guanylate cyclase activator 1A (retina)	GUCA1A	E
Guanylate kinase		E
Gustducin, alpha (taste-specific G protein)	GDCA	N
Haeme regulated inhibitor kinase		E
Haemoglobin epsilon		T
Hairless	HR	G
Haptoglobin, alpha 1	HPA1	I
Haptoglobin, alpha 2	HPA2	I
Haptoglobin, beta	HPB	I
Heat shock protein, HSP60		I
Heat shock protein, HSP70		I
Heat shock protein, HSP90		I
Heat shock protein, HSPA1		I
Heat shock protein, HSPA2		I
Hela tumor suppression gene	HTS1	G
Hemochromatosis	HFE	T
Hemopexin	HPX	I
Heparan sulfamidase		E
Heparin binding epidermal growth factor	HBEGF	G
Hepatic nuclear factor-3-beta	HNF3B	E
Hepatic nuclear factor-4-alpha	HNF4A	E
Hepatitis B virus integration site 1	HVBS1	I
Hepatitis B virus integration site 2	HVBS6	I
Hepatocyte growth factor	HGF	G
Hexosaminidase A	HEXA,TSD	E
Hexosaminidase B	HEXB	E
High mobility group protein 1	HMG1	G
High mobility group protein 2	HMG2	G
High mobility group protein C	HMGIC	G
High mobility group protein Y	HMG1Y	G
Histone family H1	H1	G
Histone family H2	H2	G
Histone family H3	H3	G
Histone family H4	H4	G
HLA-B associated transcript 1	BAT1	I
HLH transcription factor HAND1	HAND1	G
HLH transcription factor HAND2	HAND2	G
HMG-CoA lyase	HMGCL	E

HMG-CoA reductase	HMGCR	E
HMG-CoA synthase	HMGCS2	E
Holocarboxylase synthetase	HLCS	E
Holoprosencephaly 1	HPE1	G
Holoprosencephaly 2	HPE2	G
Holoprosencephaly 3	HPE3	G
Holoprosencephaly 4	HPE4	G
Homeobox (HOX) gene A1	HOXA1	G
Homeobox (HOX) gene A10	HOXA10	G
Homeobox (HOX) gene A11	HOXA11	G
Homeobox (HOX) gene A12	HOXA12	G
Homeobox (HOX) gene A13	HOXA13	G
Homeobox (HOX) gene A2	HOXA2	G
Homeobox (HOX) gene A3	HOXA3	G
Homeobox (HOX) gene A4	HOXA4	G
Homeobox (HOX) gene A5	HOXA5	G
Homeobox (HOX) gene A6	HOXA6	G
Homeobox (HOX) gene A7	HOXA7	G
Homeobox (HOX) gene A8	HOXA8	G
Homeobox (HOX) gene A9	HOXA9	G
Homeobox (HOX) gene B1	HOXB1	G
Homeobox (HOX) gene B2	HOXB2	G
Homeobox (HOX) gene B3	HOXB3	G
Homeobox (HOX) gene B4	HOXB4	G
Homeobox (HOX) gene B5	HOXB5	G
Homeobox (HOX) gene B6	HOXB6	G
Homeobox (HOX) gene B7	HOXB7	G
Homeobox (HOX) gene B8	HOXB8	G
Homeobox (HOX) gene B9	HOXB9	G
Homeobox (HOX) gene C13	HOXC13	G
Homeobox (HOX) gene C4	HOXC4	G
Homeobox (HOX) gene C8	HOXC8	G
Homeobox (HOX) gene C9	HOXC9	G
Homeobox (HOX) gene D1	HOXD1	G
Homeobox (HOX) gene D10	HOXD10	G
Homeobox (HOX) gene D12	HOXD12	G
Homeobox (HOX) gene D13	HOXD13	G
Homeobox (HOX) gene D3	HOXD3	G
Homeobox (HOX) gene D4	HOXD4	G
Homeobox (HOX) gene D8	HOXD8	G
Homeobox (HOX) gene D9	HOXD9	G
Homeobox 11	HOX11	G
Homeobox HB24	HLX1	G
Homeobox HB9	HLXB9	G
Homeobox, PROX1	PROX1	G
HSSB, replication protein		E
Human atonal gene	ATOH1	G
Human chorionic gonadotrophin, hCG	CG	G

Human placental lactogen	CSH1	G
Huntingtin	HD	T
Hypoxanthine-guanine phosphoribosyltransferase, HGPRT	HPRT	E
Hypoxia inducible factor 1	HIF1A	E
Hypoxia inducible factor 2		E
IC7 A and B		I
Iduronate 2 sulphatase	IDS	E
Ikaros gene	IKAROS	G
Immunoglobulin alpha (IgA)	IGHA	I
Immunoglobulin delta (IgD)	IGHD	I
Immunoglobulin E (IgE) responsiveness gene	IGER	I
Immunoglobulin E (IgE) serum concentration regulator gene	IGES	I
Immunoglobulin epsilon (IgE)	IGHE	I
Immunoglobulin gamma (IgG) 2	IGHG2	I
Immunoglobulin heavy mu chain	IGHM	I
Immunoglobulin J polypeptide	IGJ	I
Immunoglobulin kappa constant region	IGKC	I
Immunoglobulin kappa variable region	IGKV	I
Indian hedgehog, ihh	IHH	G
Inhibin, alpha	INHA	G
Inhibin, beta A	INHBA	G
Inhibin, beta B	INHBB	G
Inhibin, beta C	INHBC	G
Inosine monophosphate dehydrogenase, IMPDH		E
Inositol 1,4,5-triphosphate receptor 1	ITPR1	G
Inositol 1,4,5-triphosphate receptor 3	ITPR3	G
Insulin	INS	G
Insulin promotor factor 1	IPF1	G
Insulin receptor	INSR	G
Insulin receptor substrate-1	IRS1	G
Insulin-like growth factor 1	IGF1	G
Insulin-like growth factor 1 receptor	IGF1R	G
Insulin-like growth factor 2	IGF2	G
Insulin-like growth factor 2 receptor	IGF2R	G
Integrin beta 1	ITGB1	G
Integrin beta 2	ITGB2	G
Integrin beta 3	ITGB3	G
Integrin beta 4	ITGB4	G
Integrin beta 5	ITGB5	G
Integrin beta 6	ITGB6	G
Integrin beta 7	ITGB7	G
Integrin, alpha 1	ITGA1	G
Integrin, alpha 2	ITGA2	G
Integrin, alpha 3	ITGA3	G
Integrin, alpha 4	ITGA4	G

Integrin, alpha 5	ITGA5	G
Integrin, alpha 6	ITGA6	G
Integrin, alpha 7	ITGA7	G
Integrin, alpha 8	ITGA8	G
Integrin, alpha 9	ITGA9	G
Integrin, alpha M	ITGAM	G
Integrin, alpha X	ITGAX	G
Inter-alpha-trypsin inhibitor, IAT		E
Intercellular adhesion molecule 1	ICAM1	I
Intercellular adhesion molecule 2	ICAM2	I
Intercellular adhesion molecule 3	ICAM3	I
Interferon alpha	IFNA1	I
Interferon beta	IFNB	I
Interferon gamma	IFNG	I
Interferon gamma receptor 1	IFNGR1	I
Interferon gamma receptor 2	IFNGR2	I
Interferon regulatory factor 1	IRF1	I
Interferon regulatory factor 4	IRF4	I
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) 10	IL10	I
Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I
Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
IP3 kinase		E

Isocitrate dehydrogenase		E
Isovaleric acid CoA dehydrogenase	IVD	E
Janus kinase 1	JAK1	G
Janus kinase 2	JAK2	G
Janus kinase 3	JAK3	G
Kallman syndrome gene 1	KAL1	G
Kell blood group precursor	XK, KEL	T
Keratin 1	KRT1	S
Keratin 10	KRT10	S
Keratin 11	KRT11	S
Keratin 12	KRT12	S
Keratin 13	KRT13	S
Keratin 14	KRT14	S
Keratin 15	KRT15	S
Keratin 16	KRT16	S
Keratin 17	KRT17,PCHC1	S
Keratin 18	KRT18	S
Keratin 2	KRT2	S
Keratin 3	KRT3	S
Keratin 4	KRT4	S
Keratin 5	KRT5	S
Keratin 6	KRT6	S
Keratin 7	KRT7	S
Keratin 8	KRT8	S
Keratin 9	KRT9	S
Ketohexokinase	KHK	E
Kinectin	KTN1	G
Kinesin, heavy chain	KNSL1	G
Kinesin, light chain	KNS2	G
L1 cell adhesion molecule	L1CAM	N
Lactotransferrin	LTF	T
Lamin A/C	LMNA	G
Laminin 5, alpha 3	LAMA3	G
Laminin 5, beta 3	LAMB3	G
Laminin 5, gamma 2	LAMC2	G
Laminin M	LAMM	G
Laminin receptor 1	LAMR1	G
Latent transforming growth factor-beta binding protein 2	LTBP2	G
Leptin	LEP	G
Leptin receptor	LEPR	G
Leukaemia inhibitory factor	LIF	G
Leukaemia inhibitory factor receptor	LIFR	G
Leukin		I
Leukocyte-specific transcript 1	LST-1	I
Leukotriene A4 hydrolase		I
Leukotriene A4 synthase	LTA4S	E
Leukotriene B4 receptor		I

Leukotriene B4 synthase	LTB4S	E
Leukotriene C4 receptor		I
Leukotriene C4 synthase	LTC4S	E
Leukotriene D4/E4 receptor		I
LH/choriogonadotropin (CG) receptor	LHCGR	G
LIM homeobox protein 1	LHX1	G
LIM homeobox protein 2	LHX2	G
LIM homeobox protein 3	LHX3	G
LIM homeobox protein 4	LHX4	G
LIM homeobox transcription factor 1, beta	LMX1B	G
Limb girdle muscular dystrophy 1A	LGMD1A	G
Limb girdle muscular dystrophy 1B	LGMD1B	G
Limb girdle muscular dystrophy 2G	LGMD2G	G
Limb girdle muscular dystrophy 2H	LGMD2H	G
Limbic associated membrane protein	LAMP	G
LIM-domain only protein 1	LMO1	G
LIM-domain only protein 2	LMO2	G
LIM-domain only protein 3	LMO3	G
LIM-domain only protein 4	LMO4	G
Lipoma-preferred partner gene	LPP	G
Lipoprotein receptor, Low Density	LDLR	T
Lipoxygenase 12 (platelets)	LOG12	I
Lipoxygenase 5 (leukocytes)		I
Long QT-type 2 potassium channels	LQT2, KCNH2	T
Loricrin	LOR	S
Low density lipoprotein receptor-related protein precursor	LRP	T
Luteinizing hormone, beta chain	LHB	G
Lymphoblastic leukemia derived sequence 1	LYL1	I
Lymphocyte-specific protein tyrosine kinase	LCK	I
Lymphoid enhancer-binding factor	LEF-1	G
Lysosome-associated membrane protein 1	LAMP1	G
Lysosome-associated membrane protein 2	LAMP2	G
MAD (mothers against decapentaplegic, Drosophila) homologue 2	MADH2	G
MAD (mothers against decapentaplegic, Drosophila) homologue 3	MADH3	G
MAD (mothers against decapentaplegic, Drosophila) homologue 4	MADH4	G
MADS box transcription-enhancer factor 2A	MEF2A	G
MADS box transcription-enhancer factor 2B	MEF2B	G
MADS box transcription-enhancer factor 2C	MEF2C	G
MADS box transcription-enhancer factor 2D	MEF2D	G
Malate dehydrogenase, mitochondrial	MDH2	E
Malignant proliferation, eosinophil gene	MPE	I
Malonyl CoA decarboxylase		E
Malonyl CoA transferase		E
Mannosidase, alpha B lysosomal	MANB	E

Mannosidase, beta A lysosomal	MANBA	E
MAPK kinase 1	MAPKK1; MEK1	G
MAPK kinase 4	MAPKK4; MEK4;	G
	SERK1	
MAPK kinase 6	MAPKK6; MEK6	G
MAPKK kinase	MAPKKK	G
Matrix Gla protein	MGP	G
Matrix metalloproteinase 1	MMP1	E
Matrix metalloproteinase 10	MMP10	E
Matrix metalloproteinase 11	MMP11	E
Matrix metalloproteinase 12	MMP12	E
Matrix metalloproteinase 13	MMP13	E
Matrix metalloproteinase 14	MMP14	E
Matrix metalloproteinase 15	MMP15	E
Matrix metalloproteinase 16	MMP16	E
Matrix metalloproteinase 17	MMP17	E
Matrix metalloproteinase 18	MMP18	E
Matrix metalloproteinase 19	MMP19	E
Matrix metalloproteinase 2	MMP2	E
Matrix metalloproteinase 3	MMP3, STMY1	E
Matrix metalloproteinase 4	MMP4	E
Matrix metalloproteinase 5	MMP5	E
Matrix metalloproteinase 6	MMP6	E
Matrix metalloproteinase 7	MMP7	E
Matrix metalloproteinase 8	MMP8	E
Matrix metalloproteinase 9	MMP9	E
MAX-interacting protein 1	MXI1	G
MEK kinase, MEKK		E
Melanocortin 1 receptor	MC1R	T
Melanocortin 2 receptor	MC2R	T
Melanocortin 4 receptor	MC4R	T
Menin	MEN1	G
Mesoderm-specific transcript	MEST	G
Methionine adenosyltransferase	MAT1A, MAT2A	E
Methionine synthase	MTR	E
Methionine synthase reductase	MTRR	E
Methylguanine-DNA methyltransferase	MGMT	E
Methylmalonyl-CoA mutase	MUT	E
Mevalonate kinase	MVK	E
MHC Class I: A		I
MHC Class I: B		I
MHC Class I: C		I
MHC Class I: LMP-2, LMP-7		I
MHC Class I: Tap1	ABCR, TAP1	I
MHC Class II: DP	HLA-DPB1	I
MHC Class II: DQ		I
MHC Class II: DR		I
MHC Class II: Tap2	TAP2, PSF2	I



MHC Class II:Complementation group A	MHC2TA	I
MHC Class II:Complementation group B	rfxank	I
MHC Class II:Complementation group C	RFX5	I
MHC Class II:Complementation group D	RFXAP	I
Microphthalmia-associated transcription factor	MITF	G
Microsomal triglyceride transfer protein	MTP	T
Microtubule associated protein	MAP	S
Midline 1	MID1	G
Mismatch repair gene, PMSL1	PMS1	G
Mismatch repair gene, PMSL2	PMS2	G
Mitochondrial trifunctional protein, alpha subunit	HADHA	E
Mitochondrial trifunctional protein, beta subunit	HADHB	E
Mitogen-activated protein (MAP) kinase	MAPK	G
Molybdenum cofactor synthesis 1	MOCS1	E
Molybdenum cofactor synthesis 2	MOCS2	E
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Monocyte chemoattractant protein 1	MCP1	I
Motilin	MLN	G
Msh homeobox homolog 1	MSX1	G
Msh homeobox homolog 2	MSX2	G
Mucopolidoses	GNPTA	E
Mulibrey nanism	MUL	T
Multidrug resistance associated protein	MRP	G
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Muscle phosphorylase	PYGM	E
Mutated in colorectal cancers, MCC	MCC	G
MutL homolog 1	MLH1	G
MutS homolog 2	MSH2	G
MutS homolog 3	MSH3	G
Myelin protein peripheral 22	PMP22	S
Myelin protein zero	MPZ	S
Myelodysplasia syndrome 1 gene	MDS1	G
Myeloid leukemia factor-1	MLF1	I
Myocilin	MYOC	T
Myogenic factor 3	MYF3	G
Myogenic factor 4	MYF4	G
Myogenic factor 5	MYF5	G
Myomesin 1	MYOM1	S
Myomesin 2	MYOM2	S
Myosin 15	MYO15	S
Myosin 6	MYO6	S
Myosin 7A	MYO7A	S

Myosin, cardiac	MYH7	S
Myotubularin	MTM1	S
Na <sup>+</sup> , K <sup>+</sup> ATPase, alpha	ATP1A1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 1	ATP1B1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 2	ATP1B2	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 3	ATP1B3	G
Na <sup>+</sup> /H <sup>+</sup> exchanger 1	NHE1	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 2	NHE2	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 3	NHE3	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 4	NHE4	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 5	NHE5	T
N-acetylgalactosamine-6-sulfate sulfatase	GALNS	E
N-acetylglucosamine-6-sulfatase	GNS	E
N-acetylglucosaminidase, alpha	NAGLU	E
N-acetyltransferase 1	NAT1	E
N-acetyltransferase 2	NAT2	E
NADH dehydrogenase		E
NADH dehydrogenase (ubiquinone) Fe-S protein 1	NDUFS1	E
NADH dehydrogenase (ubiquinone) Fe-S protein 4	NDUFS4	E
NADH dehydrogenase (ubiquinone) flavoprotein 1	NDUFV1	E
NADH-cytochrome b5 reductase	DIA1	E
NADPH-dependent cytochrome P450 reductase	POR	E
Natural resistance-associated macrophage protein 1	NRAMP1	I
NB6		I
Necdin	NDN	G
Nephronophthisis 1	NPHP1	T
Nephronophthisis 2	NPHP2	T
Nephrosis 1	NPHS1	T
Nerve growth factor	NGF	G
Nerve growth factor receptor	NGFR	G
Neural retina-specific gene	NRL	G
Neuraminidase sialidase	NEU	T
Neuregulin	HGL	G
Neurite growth-promoting factor 2	MDK	N
Neurite inhibitory protein		N
Neuroendocrine convertase 1	NEC1, PCSK1	E
Neurofibromin 1	NF1	G
Neurofibromin 2	NF2	G
Neurofilament protein, heavy	NFH	S
Neurofilament protein, NF125	NF150	S
Neurofilament protein, NF200	NF200	S
Neurofilament protein, NF68	NF68	S
Neuronal apoptosis inhibitory protein	NAIP	I

Neuronal molecule-1		I
Neuronal molecule-1 receptor		I
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Neurotrophic tyrosine kinase receptor 1	NTRK1	G
Neurotrophin 3	NTF3 or NT3	G
Neurturin	NRTN	G
Neutral endopeptidase		E
Neutrophil cystolic factor 1	NCF1	I
Neutrophil cystolic factor 2	NCF2	I
Niacin receptor		G
Nibrin	NBS1	G
Nitric oxide synthase 1, NOS1	NOS1	E
Nitric oxide synthase 2, NOS2	NOS2	E
Nitric oxide synthase 3, NOS3	NOS3	E
Nodal	NODAL	G
Noggin	NOG	G
Norrie disease protein	NDP	G
Notch 1	NOTCH1	G
Notch 2	NOTCH2	G
Notch 3	NOTCH3	G
Notch ligand - jagged 1	JAG1, AGS	G
Nuclear factor I-kappa-B-like gene	IKBL	I
Nuclear factor kappa beta	NFKB	I
Nuclear factor of activated T cells (NFAT) complex, cytosolic	NFATC	G
Nuclear factor of activated T cells (NFAT) complex, preexisting component	NFATP	G
Nuclear mitotic apparatus protein 1	NUMA1	G
Nucleophosmin	NPM1	T
Nucleoside diphosphate kinase-A	NDPKA	E
Ocular albinism 1	OA1	S
Oculocutaneous albinism II	OCA2	S
Oligophrenin-1	OPHN1	G
Oncogene abl1	ABL1	G
Oncogene abl2		G
Oncogene akt1		G
Oncogene akt2	AKT2	G
Oncogene axl	AXL	G
Oncogene bcl2		G
Oncogene bcr/abl		G
Oncogene B-lym		G
Oncogene B-raf		G
Oncogene clk1		G
Oncogene c-myc		G
Oncogene cot		G
Oncogene crk		G

Oncogene crkl		G
Oncogene ect2		G
Oncogene ELK1	ELK1	G
Oncogene ELK2	ELK2	G
Oncogene ems1		G
Oncogene ERB		G
Oncogene ERB2		G
Oncogene ERBA		G
Oncogene ERBAL2		G
Oncogene ERG (early reponse gene)		G
Oncogene ETS1		G
Oncogene ETS2		G
Oncogene EVI1	EVI1	G
Oncogene fes		G
Oncogene fgr		G
Oncogene fos	FOS	G
Oncogene fps		G
Oncogene GLI1	GLI	G
Oncogene GLI2	GLI2	G
Oncogene GLI3	GLI3	G
Oncogene gro1		G
Oncogene gro2		G
Oncogene Ha-ras	HRAS	G
Oncogene hs1		G
Oncogene hst	FGF4	G
Oncogene int1	WNT1	G
Oncogene int2	FGF3	G
Oncogene int3	Notch4	G
Oncogene int4	WNT3	G
Oncogene jun	JUN	G
Oncogene KIT	KIT, PBT	G
Oncogene LCO	LCO	G
Oncogene l-myc		G
Oncogene lpsa		G
Oncogene lyn		G
Oncogene maf		G
Oncogene mas1		G
Oncogene mcf2		G
Oncogene mdm2	MDM2	G
Oncogene mel		G
Oncogene met	MET	G
Oncogene mos		G
Oncogene mpl		G
Oncogene MUM1	MUM1	G
Oncogene myb	MYB	G
Oncogene myc	MYC	G
Oncogene n-myc		G
Oncogene N-ras (neuroblastoma v-ras)	NRAS	G

Oncogene ovc		G
Oncogene pim1		G
Oncogene pti-1sea		G
Oncogene pvt1		G
Oncogene raf	RAF	G
Oncogene ralb		G
Oncogene rel		G
Oncogene ret	RET	G
Oncogene r-myc		G
Oncogene ros		G
Oncogene R-ras		G
Oncogene sis	PDGFB	G
Oncogene ski		G
Oncogene sno		G
Oncogene spi1		G
Oncogene src		G
Oncogene tc21		G
Oncogene TEL	ETV6	G
Oncogene tim		G
Oncogene vavtrk		G
Oncogene v-Ki-ras2	KRAS2	G
Oncogene yes		G
Oncogene yuasa		G
Oncostatin M	OSM	G
Oncostatin M receptor	OSMR	G
Orexin	OX	G
Orexin 1 receptor	OX1R	G
Orexin 2 receptor	OX2R	G
Ornithine delta-aminotransferase	OAT	E
Ornithine transcarbamoylase	OTC, NME1	E
Orthodenticle (Drosophila) homolog 1	OTX1	G
Orthodenticle (Drosophila) homolog 2	OTX2	G
Osteocalcin		S
Osteonectin	ON	G
Osteopontin	OPN	G
Osteoprotegerin	OPG	G
Otoferlin	OTOF	N
Oxytocin	OXT	N
Oxytocin receptor	OXTR	N
p21-activated kinase 3	PAK3	G
Paired box homeotic gene 1	PAX1	G
Paired box homeotic gene 2	PAX2	G
Paired box homeotic gene 3	PAX3	G
Paired box homeotic gene 6	PAX6	G
Paired box homeotic gene 7	PAX7	G
Paired box homeotic gene 8	PAX8	G
Paired-like homeodomain transcription factor 2	PITX2	G
Paired-like homeodomain transcription factor 3	PITX3	G

Palmitoyl-protein thioesterase	PPT	T
Pancreatic amylase		E
Parathyroid hormone	PTH	G
Parathyroid hormone receptor	PTHR1	G
Parathyroid hormone related-peptide	PTHrP	G
Parathyroid hormone-like hormone	PTHLH	G
Parvalbumin	PVALB	G
Patched (Drosophila) homolog, PTCH	PTCH	G
PCNA (proliferating cell nuclear antigen)		E
Peanut-like 1	PNUTL1	I
Pendrin, PDS	PDS	T
Peptidylglycine alpha-amidating monooxygenase	PAM	E
Peripherin, PRPH		S
Peroxisomal membrane protein 1	PXMP1	S
Peroxisomal membrane protein 3	PXMP3	T
Peroxisome biogenesis factor 1	PEX1	T
Peroxisome biogenesis factor 19	PEX19	T
Peroxisome biogenesis factor 6	PEX6	T
Peroxisome biogenesis factor 7	PEX7	T
Peroxisome proliferative activated receptor, alpha	PPARA	T
Peroxisome proliferative activated receptor, gamma	PPARG	T
Peroxisome receptor 1	PXR1	T
Phenylethanolamine N-methyltransferase, PNMT	PNMT	E
Phosphatase & tensin homolog	PTEN	G
Phosphate regulating gene with homologies to endopeptidases on the X chromosome	PHEX	G
Phosphatidylinositol glycan, class A (paroxysmal nocturnal hemoglobinuria)	PIGA	G
Phosphatidylinositol transfer protein	PITPN	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 1	PDNP1	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 2	PDNP2	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 3	PDNP3	G
Phosphofructokinase, liver	PFKL	E
Phosphofructokinase, muscle	PFKM	E
Phosphoglucose isomerase	GPI	E
Phosphoglycerate kinase 1	PGK1	E
Phosphoglycerate mutase 2	PGAM2	E
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I

Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C alpha		I
Phospholipase C beta		I
Phospholipase C delta	PLCD1	I
Phospholipase C epsilon		I
Phospholipase C gamma	PLCG1	I
Phosphomannomutase 1	PMM1	G
Phosphomannomutase 2	PMM2	G
Phosphomannomutase-2	PMM2	T
Phosphorylase kinase deficiency, liver	PHK	E
Phosphorylase kinase, alpha 2	PHKA2	E
Phytanoyl-CoA hydroxylase	PHYH	G
Plakophilin. 1	PKP1	T
Plasminogen	PLG	E
Plasminogen activator inhibitor 1	PAI1	E
Plasminogen activator inhibitor 2	PAI2	E
Plasminogen activator receptor, Urokinase	UPAR; PLAUR	S
Plasminogen activator, Tissue	PLAT; TPA	E
Plasminogen activator, Urokinase	UPA; PLAU	E
Platelet derived growth factor	PDGF	G
Platelet derived growth factor receptor	PDGFR	G
Plectin 1	PLEC1	T
Poly (ADP-ribose) synthetase	PARS	E
Poly(A) binding protein 2	PABP2	G
Postsynaptic density-95 protein	PSD95	N
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium inwardly-rectifying channel J11	KCNJ11	N
Potassium voltage-gated channel A1	KCNA1	N
Potassium voltage-gated channel E1	KCNE1	N
Potassium voltage-gated channel Q1	KCNQ1	N
Potassium voltage-gated channel Q2	KCNQ2	N
Potassium voltage-gated channel Q3	KCNQ3	N
Potassium voltage-gated channel Q4	KCNQ4	N
POU domain, class 1, transcription factor 1 (Pit1)	POU1F1	G
POU domain, class 3, transcription factor 4	POU3F4	G
POU domain, class 4, transcription factor 3	POU4F3	G
Pre-B-cell leukemia transcription factor 1	PBX1	G
Preproglucagon	GCG;GLP1; GLP2	G
Procollagen N-protease		E
Procollagen peptidase		E
Profibrinolysin		G
Progesterone receptor (RU486 binding receptor)	PGR	G
Prohibitin	PHB	G

Prolactin	PRL	G
Prolactin receptor	PRLR	G
Prolactin releasing hormone	PRH	G
Proliferin	PLF	G
Proline dehydrogenase	PRODH	E
Pro-melanin-concentrating hormone	PMCH	G
Promyelocytic leukemia gene	PML	G
Proopiomelanocortin	POMC	N
Prophet of Pit1	PROP1	G
Propionyl-CoA carboxylase, alpha	PCCA	E
Propionyl-CoA carboxylase, beta	PCCB	E
Prosaposin	PSAP	N
Prostaglandin (PG) D synthase, hematopoietic	PGDS	E
Prostaglandin isomerase		G
Prostaglandin-endoperoxidase synthase 2	PTGS2	G
Prostate cancer anti-metastasis gene KAI1	KAI1	G
Protease nexin 2	PN2	E
Protective protein for beta-galactosidase	PPGB	E
Protein C	PROC	I
Protein kinase A		E
Protein kinase B	PRKB	
Protein kinase C, alpha	PRKCA	E
Protein kinase C, gamma	PRKCG	E
Protein kinase DNA-activated	PRKDC	E
Protein kinase G		E
Protein phosphatase 1, regulatory (inhibitor) subunit 3	PPP1R3	E
Protein phosphatase 2, regulatory subunit A, beta isoform	PPP2R1B	E
Protein tyrosine phosphatase, non-receptor type 12	PTPN12	G
Protoporphyrinogen oxidase	PPOX	E
Pterin-4-alpha-carbinolamine	PCBD	
Purine nucleoside phosphorylase	NP	E
Purinergic receptor P1A1		N
Purinergic receptor P1A2		N
Purinergic receptor P1A3		N
Purinergic receptor P2X, 1	P2RX1	N
Purinergic receptor P2X, 2	P2RX2	N
Purinergic receptor P2X, 3	P2RX3	N
Purinergic receptor P2X, 4	P2RX4	N
Purinergic receptor P2X, 5	P2RX5	N
Purinergic receptor P2X, 6	P2RX6	N
Purinergic receptor P2X, 7	P2RX7	N
Purinergic receptor P2Y, 1	P2RY1	N
Purinergic receptor P2Y, 11	P2RY11	N
Purinergic receptor P2Y, 2	P2RY2	N
Pyrroline-5-carboxylate synthetase	PYCS	E



Pyruvate carboxylase	PC	E
Pyruvate decarboxylase	PDHA	E
Pyruvate kinase	PKLR	E
RAD51, DNA repair protein	RAD51	G
RAD52, DNA repair protein	RAD52	G
RAD54, DNA repair protein	RAD54	G
RAD55, DNA repair protein	RAD55	G
RAD57, DNA repair protein	RAD57	G
Ras-G-protein	RAS	G
Rathke pouch homeobox, RPX	RPX	G
Receptor tyrosine kinase (RTK), Nsk2	NSK2	G
Recombination activating gene 1	RAG1	G
Recombination activating gene 2	RAG2	G
Red cone pigment	RCP	S
Relaxin H1	RLN1	G
Relaxin H2	RLN2	G
Replication factor A		E
Replication factor C	RFC2	E
Retinal pigment epithelium specific protein (65kD)	RPE65	S
Retinitis pigmentosa gene 1	RP1	S
Retinitis pigmentosa gene 2	RP2	S
Retinitis pigmentosa gene 3	RP3	S
Retinitis pigmentosa gene 6	RP6	S
Retinitis pigmentosa gene 7	RP7, RDS	S
Retinoblastoma 1	RB1	G
Retinoic acid receptor, alpha	RARA	G
Retinoic acid receptor, beta	RARB	G
Retinoic acid receptor, gamma	RARG	G
Retinoid X receptor, alpha	RXRA	G
Retinoid X receptor, beta	RXRB	G
Retinoid X receptor, gamma	RXRG	G
Retinoschisis, X-linked, juvenile	RS	G
Rhabdoid tumors	SMARCB1	G
Rhodopsin	RHO	S
Ribonucleotide reductase, RRM		E
Ribosomal protein L13A	RPL13A	G
Ribosomal protein L17	RPL17	G
Ribosomal protein S19	RPS19	E
Ribosomal protein S4, X-linked	RPS4X	E
Ribosomal protein S6 kinase	RPS6KA3	E
Ribosomal protein S9	RPS9	G
RIGUI	RIGUI	G
Rod outer segment membrane protein 1	ROM1	S
Ryanodine receptor 1, skeletal	RYR1	G
SA homolog	SAH	G
Sal-like 1	SALL1	G
Secretin	SCT	T

Semaphorin A4	SEMA4	S
Semaphorin A5	SEMA5	S
Semaphorin D		S
Semaphorin E	SEMAE	S
Semaphorin F	SEMA3/F	S
Semaphorin W	SEMAW	S
Serine/threonine kinase 11	STK11	G
Serine/threonine kinase 2	STK2	G
Serotonin N-acetyltransferase	SNAT	E
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Serum amyloid A	SAA	T
Serum amyloid P	SAP	T
Sex determining region Y, SRY	SRY	G
Short stature homeobox	SHOX	G
Sialoprotein, bone	BSP	G
Signal transducer and activator of transcription 1	STAT1	G
Signal transducer and activator of transcription 2	STAT2	G
Signal transducer and activator of transcription 3	STAT3	G
Signal transducer and activator of transcription 4	STAT4	G
Signal transducer and activator of transcription 5	STAT5	G
Signaling lymphocyte activation molecule	SLAM	I
Sine oculis homeobox, drosophila, homolog 1	SIX1	G
Sine oculis homeobox, drosophila, homolog 2	SIX2	G
Sine oculis homeobox, drosophila, homolog 5	SIX5	G
Sjogren (Sjogren) syndrome antigen A1	SSA1	I
Slug protein		G
Small nuclear ribonucleoprotein polypeptide N	SNRPN	S
Smoothelin	SMTN	G
Smoothened (Drosophila) homolog	SMOH	G
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N

Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage gated, type IV, alpha polypeptide	SCN4A	N
Sodium channel, voltage gated, type V, alpha polypeptide	SCN5A	N
Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Solute carrier family 1 (amino acid transporter), member 6	SLC1A6	T
Solute carrier family 1 (glial high affinity glutamate transporter), member 3	SLC1A3	T
Solute carrier family 1 (glutamate transporter), member 1	SLC1A1	T
Solute carrier family 1 (glutamate transporter), member 2	SLC1A2	T
Solute carrier family 1 (neutral amino acid transporter), member 4	SLC1A4	T
Solute carrier family 10 (sodium/bile acid cotransporter family), member 1	SLC10A1	T
Solute carrier family 10 (sodium/bile acid cotransporter family), member 2	SLC10A2	T
Solute carrier family 12, member 1	SLC12A1	T
Solute carrier family 12, member 2	SLC12A2	T
Solute carrier family 12, member 3	SLC12A3	T
Solute carrier family 14, member 2	SLC14A2	T
Solute carrier family 15 (H <sup>+</sup> /peptide transporter, intestinal), member 1	SLC15A1	T
Solute carrier family 15 (H <sup>+</sup> /peptide transporter, kidney), member 2	SLC15A2	T
Solute carrier family 16 (monocarboxylate transporter), member 1	SLC16A1	T
Solute carrier family 16 (monocarboxylate transporter), member 7	SLC16A7	T
Solute carrier family 17, member 1	SLC17A1	T
Solute carrier family 17, member 2	SLC17A2	T
Solute carrier family 18, member 3	SLC18A3	T
Solute carrier family 19 (folate transporter), member 1	SLC19A1	T
Solute carrier family 2 (facilitated glucose transporter), member 1	SLC2A1	T
Solute carrier family 2 (facilitated glucose transporter), member 2	SLC2A2	T
Solute carrier family 2 (facilitated glucose transporter), member 3	SLC2A3	T
Solute carrier family 2 (facilitated glucose transporter), member 4	SLC2A4	T
Solute carrier family 2 (facilitated glucose	SLC2A5	T

transporter), member 5		
Solute carrier family 20, member 1	SLC20A1	T
Solute carrier family 20, member 2	SLC20A2	T
Solute carrier family 20, member 3	SLC20A3	T
Solute carrier family 21, member 2	SLC21A2	T
Solute carrier family 21, member 3	SLC21A3	T
Solute carrier family 22, member 1	SLC22A1	T
Solute carrier family 22, member 2	SLC22A2	T
Solute carrier family 22, member 5	SLC22A5	T
Solute carrier family 25, member 12	SLC25A12	T
Solute carrier family 3 (facilitated glucose transporter), member 1	SLC3A1	T
Solute carrier family 4 (anion exchanger), member 1	SLC4A1	T
Solute carrier family 4 (anion exchanger), member 2	SLC4A2	T
Solute carrier family 4 (anion exchanger), member 3	SLC4A3	T
Solute carrier family 5 (sodium/glucose transporter), member 1	SLC5A1	T
Solute carrier family 5 (sodium/glucose transporter), member 2	SLC5A2	T
Solute carrier family 5 (sodium/glucose transporter), member 5	SLC5A5	T
Solute carrier family 5, member 3	SLC5A3	T
Solute carrier family 6 (GAMMA-AMINO BUTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Solute carrier family 6 (neurotransmitter transporter, serotonin), member 4	SLC6A4	T
Solute carrier family 6, member 10	SLC6A10	T
Solute carrier family 6, member 6	SLC6A6	T
Solute carrier family 6, member 8	SLC6A8	T
Solute carrier family 7 (amino acid transporter), member 1	SLC7A1	T
Solute carrier family 7 (amino acid transporter), member 2	SLC7A2	T
Solute carrier family 7 (amino acid transporter), member 7	SLC7A7	T
Solute carrier family 8 (sodium/calcium exchanger), member 1	SLC8A1	T
Somatostatin receptor, SSTR2	SSTR2	G
Somatotrophin		G
Sonic hedgehog, SHH	SHH	G
Sorbitol dehydrogenase	SORD	E

Sorcin	SRI	T
SOS1 guanine nucleotide exchange factor	SOS1	G
Spastic paraplegia 7	SPG7	G
Spectrin alpha	SPTA1	S
Spectrin beta	SPTB	S
Sperm adhesion molecule	SPAM1	G
Sperm protamine P1	PRM1	G
Sperm protamine P2	PRM2	G
Sphingomyelinase	SMPD1	E
Spinocerebellar ataxia 8 gene	SCA8	N
Split hand/foot malformation gene	DSS1	G
SRY-box 10	SOX10	G
SRY-box 11	SOX11	G
SRY-box 3	SOX3	G
SRY-box 4	SOX4	G
SRY-box 9	SOX9	G
Stem cell factor	SCF	G
Steroid 5 alpha reductase 1	SRD5A1	E
Steroid 5 alpha reductase 2	SRD5A2	E
Steroid hormone receptor responsive DNA elements		G
Steroid sulphatase	STS	E
Steroidogenic acute regulatory protein	STAR	T
Stromal derived factor 1	SDF1	G
Succinate dehydrogenase 1	SDH1	E
Succinate dehydrogenase 2	SDH2	E
Succinate thiokinase		E
Succinic semi-aldehyde dehydrogenase	ssadh	E
Sulfamidase	SGSH	G
Sulfite oxidase	SUOX	E
Sulfonylurea receptor	SUR	G
Suppression of tumorigenicity 3 gene	ST3	G
Suppression of tumorigenicity 8 gene	ST8	G
Surfactant pulmonary-associated protein A1	SFTPA1	T
Surfactant pulmonary-associated protein A2	SFTPA2	T
Surfactant pulmonary-associated protein B	SFTPB	T
Surfactant pulmonary-associated protein C	SFTPC	T
Surfactant pulmonary-associated protein D	SFTPD	T
Surfeit 1	SURF1	G
Survival of motor neuron 1, telomeric	SMN1	T
SYK-related tyrosine kinase	SRK	I
Syndecan 1	SYND1	G
Syndecan 2	SYND2	G
Syndecan 3	SYND3	G
Syndecan 4	SYND4	G
Synovial sarcoma gene 1	SSX1	G
Synovial sarcoma gene 2	SSX2	G
Talin	TLN	G

TATA binding protein	TBP	G
TATA binding protein associated factor 2A	TAF2A	G
TATA binding protein associated factor 2C2	TAF2C2	G
TATA binding protein associated factor 2D	TAF2E	G
TATA binding protein associated factor 2F	TAF2F	G
TATA binding protein associated factor 2H	TAF2H	G
TATA binding protein associated factor 2I	TAF2I	G
TATA binding protein associated factor 2J	TAF2J	G
TATA binding protein associated factor 2K	TAF2K	G
Tau protein	MAPT	S
T-BOX 1	TBX1	G
T-BOX 2	TBX2	G
T-BOX 3	TBX3	G
T-BOX 4	TBX4	G
T-BOX 5	TBX5	G
T-BOX 6	TBX6	G
T-cell acute lymphocytic leukemia 1	TAL1	I
T-cell acute lymphocytic leukemia 2	TAL2	I
T-cell receptor, alpha	TCRA	I
T-cell receptor, delta	TCRD	I
Telomerase protein component		E
Tenascin (cytotactin)		S
Tenascin XA	TNXA	S
Terminal deoxynucleotidyltransferase, TDT		E
Testis-specific protein Y	TSPY	G
Thiolase, peroxisomal		E
Thiopurine S-methyltransferase	TPMT	E
Thrombopoietin	THPO	G
Thrombospondin	THBS1	G
Thromboxane A synthase 1	TBXAS1	I
Thromboxane A2	TXA2	I
Thromboxane A2 receptor	TBXA2R	I
Thy-1 T-cell antigen	THY1	I
Thymidylate synthase	TYMS	E
Thymopoietin	TMPO	G
Thyroglobulin	TG	G
Thyroid hormone receptor, alpha	THRA	G
Thyroid hormone receptor, beta	THRB	G
Thyroid peroxidase	TPO	G
Thyroid receptor auxiliary protein	TRAP	G
Thyroid-stimulating hormone receptor	TSHR	G
Thyroid-stimulating hormone, alpha	TSHA	G
Thyroid-stimulating hormone, beta	TSHB	G
Thyrotroph embryonic factor	TEF	G
Thyrotropin releasing hormone	TRH	G
Thyrotropin releasing hormone receptor	TRHR	G
Thyroxin-binding globulin	TBG	T
TIE receptor tyrosine kinase	TIE-1	G

Tip-associated protein	TAP	I
Tissue inhibitor of metalloproteinase 1, TIMP1	TIMP1	E
Tissue inhibitor of metalloproteinase 2, TIMP2	TIMP2	E
Tissue inhibitor of metalloproteinase 3, TIMP3	TIMP3	E
Tissue inhibitor of metalloproteinase 4, TIMP4	TIMP4	E
Tissue non-specific alkaline phosphatase TNSAP		E
Titin	TTN	S
Tocopherol (alpha) transfer protein	TTPA	T
Toll-like receptor 4	TLR4	I
Topoisomerase I		E
Topoisomerase II		E
Torticollis, keloids, cryptorchidism and renal dysplasia gene	TKCR	G
Transacylase		E
Transcobalamin 1, TCN1		T
Transcobalamin 2, TCN2	TCN2	T
Transcription factor 1, hepatic	TCF1	G
Transcription factor 2, hepatic	TCF2	G
Transcription factor 3	TCF3	G
Transcription factor binding to IGHM enhancer 3	TFE3	G
Transcription factor, TUPLE1	TUPLE1	N
Transcription termination factor, RNA polymerase 1	TTF1	G
Transcription termination factor, RNA polymerase 2	TTF2	G
Transcription termination factor, RNA polymerase 3	TTF3	G
Transferrin	TF	G
Transferrin receptor	TFRC	G
Transforming growth factor, alpha	TGFA	G
Transforming growth factor, beta 2	TGFB2	G
Transforming growth factor, beta induced	TGFBI	G
Transforming growth factor, beta receptor 2	TGFBR2	G
Transglutaminase 1	TGM1	G
Transglutaminase 2	TGM2	G
Transglutaminase 4	TGM4	G
Transketolase	TKT	E
Transketolase-like 1	TKTL1	E
Translocation in renal carcinoma on chromosome 8 gene	TRC8	G
Transthyretin	TTR	T
Treacle gene	TCOF1	G
Triosephosphate isomerase	TPI1	E
Tropomyosin 1 alpha	TPM1	S
Tropomyosin 3 (non-muscle)	TPM3	S
Troponin C		S

Troponin I	TNNI3	S
Troponin T2, cardiac	TNNT2	S
Trypsin inhibitor		E
Trypsinogen 1	TRY1	E
Trypsinogen 2	TRY2	E
Tryptophan hydroxylase	TPH	E
Tubby-like protein 1	TULP1	G
Tuberous sclerosis 1	TSC1	G
Tuberous sclerosis 2	TSC2	G
Tubulin		S
Tumor susceptibility gene 101	TSG101	G
Tumour necrosis factor (TNF) receptor associated factor 1	TRAF1	I
Tumour necrosis factor (TNF) receptor associated factor 2	TRAF2	I
Tumour necrosis factor (TNF) receptor associated factor 3	TRAF3	I
Tumour necrosis factor (TNF) receptor associated factor 4	TRAF4	I
Tumour necrosis factor (TNF) receptor associated factor 5	TRAF5	I
Tumour necrosis factor (TNF) receptor associated factor 6	TRAF6	I
Tumour necrosis factor alpha	TNFA	I
Tumour necrosis factor alpha receptor	TNFAR	I
Tumour necrosis factor beta	TNFB	I
Tumour necrosis factor beta receptor	TNFBR	I
Tumour protein p53	TP53, P53	G
Tumour protein p63	TP63	G
Tumour protein p73	TP73	G
Tumour protein, translationally-controlled 1	TPT1	G
Tumour suppressor gene DRA	DRA	I
Twist (Drosophila) homolog	TWIST	G
Tyrosinase	TYR	E
Tyrosinase-related protein 1	TYRP1	E
Tyrosine aminotransferase	TAT	E
Tyrosine hydroxylase	TH	E
Ubiquitin		G
Ubiquitin activating enzyme, E1		E
Ubiquitin B	UBB	G
Ubiquitin C	UBC	G
Ubiquitin carboxyl-terminal esterase L1	UCHL1	G
Ubiquitin fusion degeneration 1-like	UFD1L	G
Ubiquitin protein ligase E3A	UBE3A	E
UDP-glucose pyrophosphorylase		E
UDP-glucuronosyltransferase 1	ugt1d, UGT1	E
UDP-glucuronosyltransferase 2	UGT2	E
Uncoupling protein 1		T



Uncoupling protein 3	UCP3	T
Undulin 1	COL14A1	S
Uridine monophosphate kinase	UMPK	I
Uridine monophosphate synthetase	UMPS	I
Uridinediphosphate(UDP)-galactose-4-epimerase	GALE	E
Uroporphyrinogen decarboxylase	UROD	E
Uroporphyrinogen III synthase	UROS	E
Usher syndrome 2A	USH2A	S
Vascular endothelial growth factor	VEGF	G
Vasoinhibitory peptide		G
Vitamin B12-binding (R) protein		G
Vitamin D receptor	VDR	G
Vitelliform macular dystrophy, atypical gene	VMD1	T
v-myc avian myelocytomatosis viral oncogene homolog	MYC	G
Von Hippel-Lindau gene	VHL	G
Werner syndrome helicase	WRN	G
Wilms tumour gene 1	WT1	G
Wilms tumour gene 2	WT2	G
Wilms tumour gene 4	WT4	G
Winged helix nude	WHN	G
Wingless family, wnt2	WNT2	G
Wingless family, wnt4	WNT4	G
Wingless family, wnt5	WNT5	G
Wingless family, wnt7	WNT7	G
Wingless family, wnt8	WNT8	G
Wiskott-Aldrich syndrome protein	WASP, THC	I
Wnt inhibitory factor, WIF-1	WIF1	G
Wolf-Hirschhorn syndrome candidate 1 gene	WHSC1	G
Wolfram syndrome 1 gene	WFS1	S
X (inactive)-specific transcript	XIST	G
Xanthine dehydrogenase	XDH	E
Xeroderma pigmentosum, complementation group A	XPA	E
Xeroderma pigmentosum, complementation group B	XPB	E
Xeroderma pigmentosum, complementation group C	XPC	E
Xeroderma pigmentosum, complementation group D		E
Xeroderma pigmentosum, complementation group E		E
Xeroderma pigmentosum, complementation group F	XPF	E
Xeroderma pigmentosum, complementation group G	ERCC5	E
X-ray repair gene	XRCC9	G

Xylitol dehydrogenase		E
YY1 transcription factor	YY1	G
Zinc finger protein 198	ZIC198	S
Zinc finger protein 2	ZIC2	S
Zinc finger protein 3	ZIC3	S
Zinc finger protein HRX	ALL1	I
Zona pellucida glycoprotein 1	ZP1	G
Zona pellucida glycoprotein 2	ZP2	G
Zona pellucida glycoprotein 3	ZP3	G
Zona pellucida receptor tyrosine kinase	ZRK	G
Zonadhesin	ZAN	G

386. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 385.

387. A set according to claim 385 or 386 in which a minority of said probes for listed genes are absent.

388. A set according to claim 385 or 386 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.

389. A set according to claim 385 or 386 in which a limited number of probes are replaced by probes for non-listed genes.

390. A set of probes for a core group of genes according to any of claims 385 to 389 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.

391. A set according to any of claims 385 to 390 consisting of probes for members of a sub-group of the core group.

392. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.

393. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.

394. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.

395. A set according to claim 392 or 393 in which said substrate is a semiconductor microchip.

396. A set according to any preceding claim for use in a biological assay for detection of said gene variants.

- 397. A set according to any preceding claim for use in the measurement of differential gene expression levels.
- 398. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
- 399. A medical device including a set according to any of claims 385 to 397 for use in an array for detection of differential gene expression levels.
- 400. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 385) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 385 and 387 to 397 and relating the probe hybridisation pattern to said variations.
- 401. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 386) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 386 to 397 and relating the probe interaction pattern to said variations.
- 402. Use of a set or device according to any of claims 385 to 397 for the prognosis and management of patients suffering from or at risk of dysfunction, damage or disease consequent on an aberration in the processes of development or of experiencing the symptoms and consequences of dysfunction, damage or disease of the body consequent to an aberration in the processes of development.
- 403. Use of a set or device according to any of claims 385 to 397 for predicting likely therapeutic response and adverse events following therapeutic intervention.
- 404. Use of a set or device according to any of claims 385 to 397 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
- 405. Use of a set or device according to any of claims 385 to 397 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
- 406. Use of a set or device according to any of claims 385 to 397 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
- 407. Use of a set or device according to any of claims 385 to 397 for the development of new strategies of therapeutic intervention and in clinical trials.
- 408. Use of a set or device according to any of claims 385 to 397 for construction of and generation of algorithms for patient and healthcare management.
- 409. Use of a set or device according to any of claims 385 to 397 for modelling or assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations
- 410. Use of a set or device according to any of claims 385 to 397 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
- 411. Use of a set or device according to any of claims 385 to 397 for predicting optimum configuration/management of thereapeutic intervention.

412. A method according to claim 400 or 401 in which the identification of gene variants is indicative of a higher risk of developing the symptoms and consequences of dysfunction, damage or disease of the body consequent to an aberration in the processes of development for the patient or individual.
413. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop the symptoms and consequences of dysfunction, damage or disease of the body consequent to an aberration in the processes of development, which method comprises:
- i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from dysfunction, damage or disease of the body consequent to an aberration in the processes of development;
  - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the dysfunction, damage or disease of the body consequent to an aberration in the processes of development;
  - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 385 to 391;
  - iv) calculating the frequencies of these alleles in the samples from i) and ii);
  - v) comparing the frequencies of these alleles in i) and ii);
  - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing dysfunction, damage or disease of the body consequent to an aberration in the processes of development.
414. A method for assessing whether a given subject will be at risk of developing symptoms, which comprises comparing said subject's genotype with a model generated by the method of claim 413.
415. A method according to any of claims 400, 401, 413 and 414 wherein at least one step is computer-controlled.
416. An assay suitable for use in a method according to any of claims 400, 401, 413 and 414; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 385 to 491 in a biological sample.
417. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing dysfunction, damage or disease of the body consequent to an aberration in the processes of development; said kit comprising:
- i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 385 or 387 to 391 in a sample of human DNA;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing dysfunction, damage or disease of the body consequent to an aberration in the processes of development.
418. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing symptoms; said kit comprising:
- i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core group of genes as defined in any of claims 386 to 391 in an expressed-protein-containing human sample;
  - ii) reagents for use in the detection process

- iii) readout indicating the probability of a patient or individual developing dysfunction, damage or disease of the body consequent to an aberration in the processes of development.

419. A set of probes according to claim 385, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.

420. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to dysfunction, damage or disease of the skin, muscle, connective tissue or bone; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

#### KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME  
T TRANSPORT & STORAGE  
S STRUCTURAL  
I IMMUNITY  
N NERVOUS TRANSMISSION  
G GROWTH & DIFFERENTIATION

SKIN, BONE, MUSCLE GENE LIST	HUGO gene symbol	Protein function
17beta hydroxysteroid oxidoreductase		E
5,10-methylenetetrahydrofolate reductase (NADPH)	MTHFR	E
6-phosphofructo-2-kinase	PFKFB1	E
Acetoacetyl 2-CoA-thiolase	ACAT2	E
Acetylcholine receptor, nicotinic, alpha A1	CHRNA1	N
Acetylcholine receptor, nicotinic, alpha A2	CHRNA2	N
Acetylcholine receptor, nicotinic, alpha A3	CHRNA3	N
Acetylcholine receptor, nicotinic, alpha A4	CHRNA4	N
Acetylcholine receptor, nicotinic, alpha A5	CHRNA5	N
Acetylcholine receptor, nicotinic, alpha A6	CHRNA6	N
Acetylcholine receptor, nicotinic, alpha A7	CHRNA7	N
Acetylcholine receptor, nicotinic, beta 1	CHRNA8	N
Acetylcholine receptor, nicotinic, beta 2	CHRNA9	N
Acetylcholine receptor, nicotinic, beta 3	CHRNA10	N
Acetylcholine receptor, nicotinic, beta 4	CHRNA11	N
Acetylcholine receptor, nicotinic, epsilon	CHRNA12	N
Acetylcholine receptor, nicotinic, gamma	CHRNA13	N
Acetylcholinesterase	ACHE	E
Achromatopsia 2	ACHM2	S
Acid phosphatase 2, lysosomal	ACP2	E

Actin, alpha, cardiac	ACTC	S
Actin, alpha, skeletal	ACTA1	S
Actin, alpha, smooth, aortic	ACTA2	S
Actin, beta	ACTB	S
Actin, gamma 2	ACTG2	S
Activin		G
Acyl CoA dehydrogenase, short chain	ACADS	E
Acyl-CoA thioesterase		E
Adaptin, beta 3A	ADTB3A	T
Adducin, alpha	ADD1	S
Adducin, beta	ADD2	S
Adenosine deaminase	ADA	E
Adenosine monophosphate deaminase	AMPD	E
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N
Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenyl cyclase		N
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adenylate kinase	AK1	E
Adenylosuccinate lyase	ADSL	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
Adrenocorticotrophic hormone (ACTH) receptor	ACTHR	G
Adrenoleukodystrophy gene	ALD	E
Alanine aminotransferase		T
Alanine-glyoxylate aminotransferase	AGXT	E
Albumin, ALB	ALB	T
Alcohol dehydrogenase 1	ADH1	E
Alcohol dehydrogenase 2	ADH2	E
Alcohol dehydrogenase 3	ADH3	E
Alcohol dehydrogenase 4	ADH4	E
Alcohol dehydrogenase 5	ADH5	E
Alcohol dehydrogenase 6	ADH6	E
Alcohol dehydrogenase 7	ADH7	E
Aldehyde dehydrogenase 1	ALDH1	E

Aldehyde dehydrogenase 10	ALDH10	E
Aldehyde dehydrogenase 2	ALDH2	E
Aldehyde dehydrogenase 5	ALDH5	E
Aldehyde dehydrogenase 6	ALDH6	E
Aldehyde dehydrogenase 7	ALDH7	E
Aldolase A	ALDOA	E
Aldolase B	ALDOB	E
Aldolase C	ALDOC	E
Aldosterone receptor	MLR	G
Alkaline phosphatase, liver/bone/kidney	ALPL	T
Alkaptonuria gene	AKU	G
Alkylglycerone phosphate synthase	AGPS	E
alpha tectorin	TECTA	G
alpha thalassemia gene	ATRX	N
alpha1-antichymotrypsin	AACT	E
alpha1-antitrypsin	PI	E
alpha2-antiplasmin	PLI	E
alpha-actinin 2	ACTN2	G
alpha-actinin 3	ACTN3	G
alpha-Galactosidase A	GLA	E
Alpha-galactosidase B, GALB	NAGA	E
alpha-synuclein	SNCA	N
Amelogenin	AMELX	S
Aminopeptidase P	XPNPEP2	E
Amphiregulin	AREG	G
Amylo-1,6-glucosidase	AGL	E
Amyloid beta A4 precursor protein	APP	N
Amyloid beta A4 precursor-like protein	APLP	N
Androgen binding protein	ABP	T
Androgen receptor	AR	G
Angiopoietin 1	ANGPT1	G
Angiopoietin 2	ANGPT2	G
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensinogen	AGT	E
Antidiuretic hormone receptor	ADHR	T
Anti-Mullerian hormone	AMH	G
Apolipoprotein A 4	APOA4	T
Apolipoprotein A I	APOA1	T
Apolipoprotein A II	APOA2	T
Apolipoprotein B	APOB	T
Apolipoprotein C1	APOC1	T
Apolipoprotein C2	APOC2	T
Apolipoprotein C3	APOC3	T
Apolipoprotein D	APOD	T
Apolipoprotein E	APOE	T
Apolipoprotein H	APOH	T
Arginine vasopressin	AVP	N
Arginine vasopressin receptor 1A	AVPR1A	N

Arginine vasopressin receptor 1B	AVPR1B	N
Arginine vasopressin receptor 2	AVPR2	N
Arrestin	SAG	S
Aryl hydrocarbon receptor nuclear translocator	ARNT	T
Arylsulfatase A	ARSA	E
Arylsulfatase B	ARSB	E
Arylsulfatase C	ARSC1	E
Arylsulfatase D	ARSD	E
Arylsulfatase E	ARSE	E
Arylsulfatase F	ARSF	E
Aspartate receptor		N
Aspartoacylase	ASPA	E
Aspartylglucosaminidase	AGA	E
Ataxia telangiectasia complementation group D	ATD, ATDC	G
Ataxia telangiectasia gene, AT	ATM	G
ATP cobalamin adenosyltransferase		E
ATP sulphurylase	atpsk2	E
ATP/ADP translocase		E
Attractin		I
Autoimmune regulator, AIRE	AIRE	I
BCL2-related protein A1	BCL2A1	G
Benzodiazepine receptor		N
Bestrophin	VMD2	T
beta 2 microglobulin	B2M	I
beta-endorphin receptor		N
beta-galactosidase	GLB1	E
beta-Glucuronidase	GUSB	E
beta-synuclein	SNCB	N
Bilirubin UDP-glucuronosyltransferase		E
Bloom syndrome protein	BLM	G
Blue cone pigment	BCP	S
Bone morphogenetic protein, BMP1	BMP1	G
Bone morphogenetic protein, BMP2	BMP2	G
Bone morphogenetic protein, BMP3	BMP3	G
Bone morphogenetic protein, BMP4	BMP4	G
Bone morphogenetic protein, BMP5	BMP5	G
Bone morphogenetic protein, BMP6	BMP6	G
Bone morphogenetic protein, BMP7	BMP7	G
Bone morphogenetic protein, BMP8	BMP8	G
Bradykinin receptor B1		I
Bradykinin receptor B2		I
Branched chain aminotransferase 1, cytosolic	BCAT1	E
Branched chain aminotransferase 2, mitochondrial	BCAT2	E
Breast cancer, ductal, 1	BRCD1	G
Breast cancer, ductal, 2	BRCD2	G



Butyrylcholinesterase	BCHE	E
Ca(2+) transporting ATPase, fast twitch	ATP2A1	T
Ca(2+) transporting ATPase, slow twitch	ATP2A2	T
Cadherin E	CDH1	G
Cadherin EP		G
Cadherin N	CDH2	G
Cadherin P	CDH3	G
Calbindin 1	CALB1	G
Calbindin D9K	CALB3	G
Calcitonin receptor /Calcitonin gene-related peptide receptor	CALCR	N
Calcitonin/Calcitonin gene-related peptide alpha	CALCA	N
Calcium channel, voltage-dependent, L type, alpha 1S subunit	CACNA1S	N
Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	CACNA1A	N
Calmodulin 1	CALM1	G
Calmodulin 2	CALM2	G
Calmodulin 3	CALM3	G
Calnexin	CANX	G
Calpain	CAPN, CAPN3	E
Cannabinoid receptor	CNR1	N
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Carnitine acetyltransferase	CRAT	E
Carnitine acylcarnitine translocase	CACT	E
Carnitine palmitoyltransferase I	CPT1A	E
Carnitine palmitoyltransferase II	CPT2	E
Carnitine transporter protein	CDSP, SCD	T
Cartilage oligomeric matrix protein	COMP, EDM1, PSACH	N
Cartilage-hair hypoplasia gene	CHH	N
Catenin, beta	CTNNB1	G
Cathepsin K	CTSK	E
Caveolin 3	CAV3	E
CD1	CD1	I
CD4	CD4	I
Ceroid lipofuscinosis neuronal 3	CLN3	N
Ceruloplasmin precursor	CP	E
Chemokine MCAF	MCAF	I
Chloride channel 1, skeletal muscle	CLCN1	S
Cholecystokinin	CCK	N
Cholecystokinin B receptor	CCKBR	N
Cholesterol ester hydroxylase		E
Choline acetyltransferase	CHAT	E

Choroideremia gene	CHM	S
Citrate synthase		E
Clathrin		T
Cleft palate gene	CPX	G
Cockayne syndrome gene, CKN1	CKN1	G
Coenzyme Q (CoQ)/ubiquinone		E
Collagen I alpha 1	COL1A1	S
Collagen I alpha 2	COL1A2	S
Collagen II alpha 1	COL2A1	S
Collagen III alpha 1	COL3A1	S
Collagen IV alpha 1	COL4A1	S
Collagen IV alpha 2	COL4A2	S
Collagen IV alpha 3	COL4A3	S
Collagen IV alpha 4	COL4A4	S
Collagen IV alpha 5	COL4A5	S
Collagen IV alpha 6	COL4A6	S
Collagen IX alpha 2	COL9A2, EDM2	S
Collagen IX alpha 3	COL9A3	S
Collagen receptor	COLR	S
Collagen V alpha 1	COL5A1	S
Collagen V alpha 2	COL5A2	S
Collagen VI alpha 1	COL6A1	S
Collagen VI alpha 2	COL6A2	S
Collagen VI alpha 3	COL6A3	S
Collagen VII alpha 1	COL7A1	S
Collagen X alpha 1	COL10A1	S
Collagen X alpha 1	COL11A1	S
Collagen XI alpha 2	COL11A2	S
Collagen XVII alpha 1	COL17A1	S
Collagenic-like tail subunit of asymmetric acetylcholinesterase	COLQ	E
Collapsin		G
Colony-stimulating factor 1	CSF1	G
Colony-stimulating factor 1 receptor	CSF1R	G
Colony-stimulating factor 2	CSF2	G
Colony-stimulating factor 2 alpha receptor	CSF2RA	G
Colony-stimulating factor 2 beta receptor	CSF2RB	G
Colony-stimulating factor 3	CSF3	G
Colony-stimulating factor 3 receptor	CSF3R	G
Complement component C1 inhibitor	C1NH	I
Complement component C1qa	C1QA	I
Complement component C1qb	C1QB	I
Complement component C1qg	C1QG	I
Complement component C1r	C1R	I
Complement component C1s	C1S	I
Complement component C2	C2	I
Complement component C3	C3	I
Complement component C4A	C4A	I

Complement component C4B	C4B	I
Complement component C5	C5	I
Complement component C6	C6	I
Complement component C7	C7	I
Complement component C8	C8B	I
Complement component C9	C9	I
Complement component receptor 1	CR1	I
Complement component receptor 2	CR2	I
Complement component receptor 3	CR3	I
Complex I		E
Complex II		E
Complex III		E
Complex III		E
Complex V	MTATP6	E
Cone-rod homeobox-containing gene	CRX	G
Coproporphyrinogen oxidase	CPO	E
Core-binding factor, alpha 1	CBFA1	G
Core-binding factor, alpha 2	CBFA2	G
Core-binding factor, beta	CBFB	G
Corticosteroid binding globulin	CBG	N
Cortico-steroid binding protein		T
Corticotrophin-releasing hormone	CRH	T
Corticotrophin-releasing hormone receptor	CRHR1	T
Cortisol receptor		I
C-reactive protein CRP		I
Creatine kinase – B and m	CKBE	E
Creb binding protein	CREBBP	G
Crystallin, alpha A	CRYAA	S
Crystallin, alpha B	CRYAB	S
Crystallin, beta B2	CRYBB2	S
Crystallin, gamma A	CRYGA	S
c-src tyrosine kinase	CSK	G
Cu <sup>2+</sup> transporting ATPase alpha polypeptide	ATP7A	E
Cu <sup>2+</sup> transporting ATPase beta polypeptide	ATP7B	E
Cyclic AMP response element binding protein	CREB	G
Cyclic AMP-dependent protein kinase	PKA	E
Cyclic nucleotide phosphodiesterase 1B	PDE1B	E
Cyclic nucleotide phosphodiesterase 1B1	PDE1B1	E
Cyclic nucleotide phosphodiesterase 2A3	PDE2A3	E
Cyclic nucleotide phosphodiesterase 3A	PDE3A	E
Cyclic nucleotide phosphodiesterase 3B	PDE3B	E
Cyclic nucleotide phosphodiesterase 4A	PDE4A	E
Cyclic nucleotide phosphodiesterase 4C	PDE4C	E
Cyclic nucleotide phosphodiesterase 5A	PDE5A	E
Cyclic nucleotide phosphodiesterase 6A	PDE6A	E
Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E
Cyclic nucleotide phosphodiesterase 8	PDE8	E

Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclin-dependent kinase 2	CDK2	G
Cyclin-dependent kinase inhibitor 1C (P57, KIP2)	CDKN1C	G
Cyclin-dependent kinase inhibitor 2A (p16)	CDKN2A	G
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
CYP11A1	CYP11A1	E
CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E
CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E
CYP2C18	CYP2C18	E
CYP2C19	CYP2C19	E
CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E
CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E
CYP2F1	CYP2F1	E
CYP2J2	CYP2J2	E
CYP3A3	CYP3A3	E
CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystathionase	CTH	E
Cystathione beta synthase	CBS	E
Cystic fibrosis transmembrane conductance	CFTR	N

regulator, CFTR		
Cystinosis	CTNS	T
Cytidine deaminase	CDA	E
Cytidine-5-prime-triphosphate synthetase	CTPS	E
Cytochrome a		E
Cytochrome b-245 alpha	CYBA	E
Cytochrome b-245 beta	CYBB	E
Cytochrome b-5	CYB5	E
Cytochrome c		E
Cytochrome c oxidase, MTCO		E
Cytokine-suppressive antiinflammatory drug-binding protein 1	CSBP1	I
Cytokine-suppressive antiinflammatory drug-binding protein 2	CSBP2	I
DAX1 nuclear receptor	DAX1	I
Deafness dystonia peptide	DDP	N
Delta 4-5 alpha-reductase		E
Delta aminolevulinate dehydratase	ALAD	E
Delta(4)-3-oxosteroid 5-beta-reductase		E
Delta-7-dehydrocholesterol reductase	DHCR7	E
Dentin sialophosphoprotein	DSPP	G
Desmin	DES	S
DHEA sulfotransferase	STD	E
Diastrophic dysplasia sulfate transporter	DTD	T
Dihydrolipoamide dehydrogenase	DLD	N
Dihydroxyacetonephosphate acyltransferase	DHAPAT	E
DNA damage binding protein, DDB1	DDB1	S
DNA damage binding protein, DDB2	DDB2	S
DNA methyltransferase	DNMT	E
DNA-damage-inducible transcript 3	DDIT3	S
Dopamine receptors D1	DRD1	N
Dopamine receptors D2	DRD2	N
Dopamine receptors D3	DRD3	N
Dopamine receptors D4	DRD4	N
Dopamine receptors D5	DRD5	N
Dynamitin	DNM1	G
Dynorphin receptor		N
Dyskerin	DKC1	S
Dystonia 1	DYT1	S
Dystonia 3	DYT3	S
Dystonia 6	DYT6	S
Dystonia 7	DYT7	S
Dystrophia myotonica	DM, DMPK	E
Dystrophia myotonica, atypical	DM2	E
Dystrophin	DMD	S
Dystrophin-associated glycoprotein 35kD, SCGD	SGCD	S
Dystrophin-associated glycoprotein 35kD,	SGCG	S

<b>SGSG</b>		
Dystrophin-associated glycoprotein 43kD	SGCB	S
Dystrophin-associated glycoprotein 50kD	SGCA	S
Ectodermal Dysplasia 1 gene	ED1	S
Elastase 1	ELAS1	E
Elastase 2	ELAS2	E
Elastin	ELN	S
Electron-transferring-flavoprotein alpha	ETFA	T
Electron-transferring-flavoprotein beta	ETFB	T
Electron-transferring flavoprotein dehydrogenase	ETFDH	E
Emerin	EMD	T
Endocardial fibroelastosis 2 gene	EFE2	S
Endometrial bleeding-associated factor	EBAF	G
Endothelin 1	EDN1	N
Endothelin 2	EDN2	N
Endothelin 3	EDN3	N
Endothelin converting enzyme	ECE1	N
Endothelin receptor type A	EDNRA	N
Endothelin receptor type B	EDNRB	N
Engrailed-1	EN1	G
Engrailed-2	EN2	G
Enolase	ENO1	E
Enoyl CoA hydratase		E
Enoyl CoA isomerase		E
Enoyl CoA reductase		E
Enterokinase	PRSS7, ENTK	E
Ephrin receptor tyrosine kinase A	EPHA	G
Ephrin receptor tyrosine kinase B	EPHB	G
Epidermal growth factor	EGF	G
Epidermal growth factor receptor	EGFR	G
Erythrocyte membrane protein band 4.1	EPB41	S
Erythropoietin	EPO	I
Erythropoietin receptor	EPOR	I
Estrogen receptor	ESR	G
Exostosin 1	EXT1	S
Exostosin 2	EXT2	S
Exostosin 3	EXT3	S
Eye colour gene 3 (brown)	EYCL3	S
Eyes absent 1	EYA1	G
Faciogenital dysplasia	FGD1, FGDY	T
Factor 1 (No. one)	F1	I
Factor B, properdin		I
Factor D		I
Factor H	HF1	I
Factor X	F10	I
Fanconi anemia, complementation group A	FANCA	T
Fanconi anemia, complementation group C	FANCC	T

Fanconi anemia, complementation group D	FANCD	T
Fc fragment of IgG, high affinity IA, receptor for	FCGR1A	G
Fc fragment of IgG, low affinity IIa, receptor for (CD32)	FCGR2A	G
Ferritin, H subunit		T
Ferritin, L subunit	FTL	T
Fibrillin 1	FBN1	G
Fibrillin 2	FBN2	G
Fibrinogen alpha	FGA	S
Fibrinogen beta	FGB	S
Fibrinogen gamma	FGG	S
Fibroblast growth factor	FGF1	G
Fibroblast growth factor receptor 1	FGFR1	G
Fibroblast growth factor receptor 2	FGFR2	G
Fibroblast growth factor receptor 3	FGFR3	G
Fibronectin precursor	FN1	G
Flightless-II, Drosophila homolog of	FLII	G
Folic acid receptor	FOLR	G
Follicle stimulating hormone receptor	FSHR, ODG1	G
Follicle stimulating hormone, FSH	FSHB	G
Forkhead transcription factor 10	FKHL10	G
Forkhead transcription factor 14	FKHL14	G
Forkhead transcription factor 7	FKHL7	G
Fragile site, folic acid type, rare, fra(X) A	FRAXA	N
Frataxin	FRDA	G
Fringe secreted protein, lunatic	LFNG	G
Fringe secreted protein, manic	MFNG	G
Fringe secreted protein, radical	RFNG	G
Fructose-1,6-diphosphatase	FBP1	E
Fucosidase alpha-L-1	FUCA1	E
Fucosidase alpha-L-2		E
Fukuyama type congenital muscular dystrophy	FCMD	G
Fumarase	FH	E
GABA receptor, alpha 1	GABRA1	N
GABA receptor, alpha 2	GABRA2	N
GABA receptor, alpha 3	GABRA3	N
GABA receptor, alpha 4	GABRA4	N
GABA receptor, alpha 5	GABRA5	N
GABA receptor, alpha 6	GABRA6	N
GABA receptor, beta 1	GABRB1	N
GABA receptor, beta 2	GABRB2	N
GABA receptor, beta 3	GABRB3	N
GABA receptor, gamma 1	GABRG1	N
GABA receptor, gamma 2	GABRG2	N
GABA receptor, gamma 3	GABRG3	N
Galactocerebrosidase	GALC	E

Galactokinase	GALK1	E
Galactose 1-phosphate uridyl-transferase	GALT	E
Gamma-glutamyl carboxylase	GGCX	T
Gap junction protein alpha 3	GJA3	T
Gap junction protein alpha 8	GJA8	T
Gap junction protein beta 3	GJB3	T
Gastrointestinal tumor-associated antigen 1	GA733	I
Gastrulation brain homeobox 2	GBX2	G
Glucosidase, acid alpha	GAA	E
Glucosidase, acid beta	GBA	E
Glutamate receptor 1	GLUR1	N
Glutamate receptor 2	GLUR2	N
Glutamate receptor 3	GLUR3	N
Glutamate receptor 4	GLUR4	N
Glutamate receptor 5	GLUR5	N
Glutamate receptor 6	GLUR6	N
Glutamate receptor 7	GLUR7	N
Glutamate receptor, ionotropic, NMDA 1	NMDAR1	N
Glutamate receptor, ionotropic, NMDA 2A	NMDAR2A	N
Glutamate receptor, ionotropic, NMDA 2B	NMDAR2B	N
Glutamate receptor, ionotropic, NMDA 2C	NMDAR2C	N
Glutamate receptor, ionotropic, NMDA 2D	NMDAR2D	N
Glutathione	GSH	T
Glutathione peroxidase, GPX1	GPX1	E
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
Glycerol kinase	GK	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
Glycine receptor, alpha	GLRA2	N
Glycine receptor, beta		N
Glycine transporter	GLYT	N
Glycogen phosphorylase	PYGL	E
Glycosyltransferases, ABO blood group	ABO	E
GM2 ganglioside activator protein, GM2A	GM2A	E
Green cone pigment	GCP	S
Growth arrest-specific homeobox	GAX	G
Growth factor receptor-bound protein 2	GRB2	G
Growth hormone 1	GH1	G
Growth hormone 2 (placental)	GH2	G
Growth hormone receptor	GHR	G
Growth hormone releasing hormone (GHRH)	GHRH	G
Growth hormone releasing hormone receptor	GHRHR	G
Growth/differentiation factor 5	GDF5	G
GTP cylcohydrolase 1	GCH1	G
GTPase-activating protein, GAP	RASA1	G
Guanidinoacetate N-methyltransferase	GAMT	E



Guanine nucleotide-binding protein, alpha activating activity polypeptide, GNAO	GNAO1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 1, GNAI1	GNAI1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 2, GNAI2	GNAI2	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 3, GNAI3	GNAI3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS1	GNAS1	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS2	GNAS2	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS3	GNAS3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS4	GNAS4	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT1	GNAT1	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT2	GNAT2	N
Guanine nucleotide-binding protein, beta polypeptide 3	GNB3	N
Guanine nucleotide-binding protein, gamma polypeptide 5	GNG5	N
Guanine nucleotide-binding protein, q polypeptide	GNAQ	N
Guanylate cyclase 2D, membrane (retina-specific)	GUCY2D	E
Guanylate cyclase activator 1A (retina)	GUCA1A	E
H(+), K(+) - ATPase	ATP4B	N
Haeme regulated inhibitor kinase		E
Haemoglobin alpha 1	HBA1	T
Haemoglobin alpha 2	HBA2	T
Haemoglobin beta	HBB	T
Haemoglobin delta	HBD	T
Haemoglobin gamma A	HBG1	T
Haemoglobin gamma B	HBG2	T
Haemoglobin gamma G	HBGG	T
Hairless	HR	G
Heat shock protein, HSP60		I
Heat shock protein, HSP70		I
Heat shock protein, HSP90		I
Heat shock protein, HSPA1		I
Heat shock protein, HSPA2		I
Heparan sulfamidase		E
Heparin binding epidermal growth factor	HBEGF	G
Heparin Cofactor II	HCF2	I
Hepatocyte growth factor	HGF	G

Hermansky-pudlak syndrome gene	HPS	T
Hexokinase 2	HK2	E
Hexosaminidase A	HEXA,TSD	E
Hexosaminidase B	HEXB	E
Histamine receptors, H1		N
Histamine receptors, H2		N
Histamine receptors, H3		N
HLA-B associated transcript 1	BAT1	I
Holocarboxylase synthetase	HLCS	E
Holoprosencephaly 1	HPE1	G
Holoprosencephaly 2	HPE2	G
Holoprosencephaly 3	HPE3	G
Holoprosencephaly 4	HPE4	G
Homeobox (HOX) gene A1	HOXA1	G
Homeobox (HOX) gene A10	HOXA10	G
Homeobox (HOX) gene A11	HOXA11	G
Homeobox (HOX) gene A12	HOXA12	G
Homeobox (HOX) gene A13	HOXA13	G
Homeobox (HOX) gene A2	HOXA2	G
Homeobox (HOX) gene A3	HOXA3	G
Homeobox (HOX) gene A4	HOXA4	G
Homeobox (HOX) gene A5	HOXA5	G
Homeobox (HOX) gene A6	HOXA6	G
Homeobox (HOX) gene A7	HOXA7	G
Homeobox (HOX) gene A8	HOXA8	G
Homeobox (HOX) gene A9	HOXA9	G
Homeobox (HOX) gene B1	HOXB1	G
Homeobox (HOX) gene B2	HOXB2	G
Homeobox (HOX) gene B3	HOXB3	G
Homeobox (HOX) gene B4	HOXB4	G
Homeobox (HOX) gene B5	HOXB5	G
Homeobox (HOX) gene B6	HOXB6	G
Homeobox (HOX) gene B7	HOXB7	G
Homeobox (HOX) gene B8	HOXB8	G
Homeobox (HOX) gene B9	HOXB9	G
Homeobox (HOX) gene C13	HOXC13	G
Homeobox (HOX) gene C4	HOXC4	G
Homeobox (HOX) gene C8	HOXC8	G
Homeobox (HOX) gene C9	HOXC9	G
Homeobox (HOX) gene D1	HOXD1	G
Homeobox (HOX) gene D10	HOXD10	G
Homeobox (HOX) gene D12	HOXD12	G
Homeobox (HOX) gene D13	HOXD13	G
Homeobox (HOX) gene D3	HOXD3	G
Homeobox (HOX) gene D4	HOXD4	G
Homeobox (HOX) gene D8	HOXD8	G
Homeobox (HOX) gene D9	HOXD9	G
Homeobox 11	HOX11	G

Homeobox HB24	HLX1	G
Homeobox HB9	HLXB9	G
Homeobox, PROX1	PROX1	G
Homogentisate 1,2 dioxygenase	HGD	E
Human placental lactogen	CSH1	G
Hypoxia inducible factor 1	HIF1A	E
Hypoxia inducible factor 2		E
IC7 A and B		I
Immunoglobulin E (IgE) responsiveness gene	IGER	I
Indian hedgehog, ihh	IHH	G
Inhibin, alpha	INHA	G
Inhibin, beta A	INHBA	G
Inhibin, beta B	INHBB	G
Inhibin, beta C	INHBC	G
Inositol 1,4,5-triphosphate receptor 3	ITPR3	G
Insulin promotor factor 1	IPF1	G
Insulin-like growth factor 1	IGF1	G
Insulin-like growth factor 1 receptor	IGF1R	G
Insulin-like growth factor 2	IGF2	G
Insulin-like growth factor 2 receptor	IGF2R	G
Integrin beta 1	ITGB1	G
Integrin beta 3	ITGB3	G
Integrin beta 4	ITGB4	G
Integrin, alpha 5	ITGA5	G
Integrin, alpha 7	ITGA7	G
Inter-alpha-trypsin inhibitor, IATI		E
Interferon alpha	IFNA1	I
Interferon beta	IFNB	I
Interferon gamma	IFNG	I
Interferon gamma receptor 1	IFNGR1	I
Interferon gamma receptor 2	IFNGR2	I
Interferon regulatory factor 1	IRF1	I
Interferon regulatory factor 4	IRF4	I
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) 10	IL10	I
Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I
Interleukin(IL) 3	IL3	I

Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
Isocitrate dehydrogenase		E
Kallman syndrome gene 1	KAL1	G
Keratin 1	KRT1	S
Keratin 10	KRT10	S
Keratin 11	KRT11	S
Keratin 12	KRT12	S
Keratin 13	KRT13	S
Keratin 14	KRT14	S
Keratin 15	KRT15	S
Keratin 16	KRT16	S
Keratin 17	KRT17,PCHC1	S
Keratin 18	KRT18	S
Keratin 2	KRT2	S
Keratin 3	KRT3	S
Keratin 4	KRT4	S
Keratin 5	KRT5	S
Keratin 6	KRT6	S
Keratin 7	KRT7	S
Keratin 8	KRT8	S
Keratin 9	KRT9	S
Keratin, hair acidic 1	KRTHA1	S
Keratin, hair basic 2	KRTHB1	S
Keratin, hair basic 6	KRTHB6	S
Kininogen, High molecular weight	KNG	I
Lactate dehydrogenase, A	LDHA	E
Lactate dehydrogenase, B	LDHB	E
Lamin A/C	LMNA	G
Laminin 5, alpha 3	LAMA3	G
Laminin 5, beta 3	LAMB3	G
Laminin 5, gamma 2	LAMC2	G
Laminin M	LAMM	G
Laminin receptor 1	LAMR1	G
Latent transforming growth factor-beta binding protein 2	LTBP2	G

Leukocyte-specific transcript 1	LST-1	I
Leukotriene A4 hydrolase		I
Leukotriene A4 synthase	LTA4S	E
Leukotriene B4 receptor		I
Leukotriene B4 synthase	LTB4S	E
Leukotriene C4 receptor		I
Leukotriene C4 synthase	LTC4S	E
LIM homeobox transcription factor 1, beta	LMX1B	G
Limb girdle muscular dystrophy 1A	LGMD1A	G
Limb girdle muscular dystrophy 1B	LGMD1B	G
Limb girdle muscular dystrophy 2G	LGMD2G	G
Limb girdle muscular dystrophy 2H	LGMD2H	G
Limbic associated membrane protein	LAMP	G
Lipoprotein receptor, Low Density	LDLR	T
Lipoxygenase 12 (platelets)	LOG12	I
Loricrin	LOR	S
Low density lipoprotein receptor-related protein precursor	LRP	T
Luteinizing hormone-releasing hormone		N
Luteinizing hormone-releasing hormone receptor		N
lymphotoxin		I
Lysosome-associated membrane protein 1	LAMP1	G
Lysosome-associated membrane protein 2	LAMP2	G
Lysozyme	LYZ	I
Lysyl hydroxylase	PLOD	E
Lysyl oxidase	LOX	E
Macrophage activating factor	MAF	I
Macrophage inflammatory protein-1	MIP1	I
Macrophage inflammatory protein-1 receptor		I
Macrophage inflammatory protein-2	MIP2	I
Macrophage inflammatory protein-2 receptor		I
MADS box transcription-enhancer factor 2A	MEF2A	G
MADS box transcription-enhancer factor 2B	MEF2B	G
MADS box transcription-enhancer factor 2C	MEF2C	G
MADS box transcription-enhancer factor 2D	MEF2D	G
Mannose binding protein	MBP	I
Mannosidase, alpha B lysosomal	MANB	E
Mannosidase, beta A lysosomal	MANBA	E
Marenostrin	MEFV	T
Matrix Gla protein	MGP	G
Matrix metalloproteinase 1	MMP1	E
Matrix metalloproteinase 10	MMP10	E
Matrix metalloproteinase 11	MMP11	E
Matrix metalloproteinase 12	MMP12	E
Matrix metalloproteinase 13	MMP13	E
Matrix metalloproteinase 14	MMP14	E
Matrix metalloproteinase 15	MMP15	E

Matrix metalloproteinase 16	MMP16	E
Matrix metalloproteinase 17	MMP17	E
Matrix metalloproteinase 18	MMP18	E
Matrix metalloproteinase 19	MMP19	E
Matrix metalloproteinase 2	MMP2	E
Matrix metalloproteinase 3	MMP3, STMY1	E
Matrix metalloproteinase 4	MMP4	E
Matrix metalloproteinase 5	MMP5	E
Matrix metalloproteinase 6	MMP6	E
Matrix metalloproteinase 7	MMP7	E
Matrix metalloproteinase 8	MMP8	E
Matrix metalloproteinase 9	MMP9	E
MEK kinase, MEKK		E
Melanocortin 1 receptor	MC1R	T
Melanocortin 2 receptor	MC2R	T
Melanocortin 4 receptor	MC4R	T
Mesoderm-specific transcript	MEST	G
Methylguanine-DNA methyltransferase	MGMT	E
Methylmalonyl-CoA mutase	MUT	E
Mevalonate kinase	MVK	E
MHC Class I: A		I
MHC Class I: B		I
MHC Class I: C		I
MHC Class I: LMP-2, LMP-7		I
MHC Class I: Tap1	ABCR, TAP1	I
MHC Class II: DP	HLA-DPB1	I
MHC Class II: DQ		I
MHC Class II: DR		I
MHC Class II: Tap2	TAP2, PSF2	I
MHC Class II:Complementation group A	MHC2TA	I
MHC Class II:Complementation group B	rfxank	I
MHC Class II:Complementation group C	RFX5	I
MHC Class II:Complementation group D	RFXAP	I
Microphthalmia-associated transcription factor	MITF	G
Midline 1	MID1	G
Mitochondrial trifunctional protein, alpha subunit	HADHA	E
Mitochondrial trifunctional protein, beta subunit	HADHB	E
Moesin, MSN		S
Molybdenum cofactor synthesis 1	MOCS1	E
Molybdenum cofactor synthesis 2	MOCS2	E
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Monocyte chemoattractant protein 1	MCP1	I
Mucopolidoses	GNPTA	E
Mulibrey nanism	MUL	T

Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Muscle phosphorylase	PYGM	E
Mutated in colorectal cancers, MCC	MCC	G
MutS homolog 3	MSH3	G
Myeloperoxidase	MPO	I
Myocilin	MYOC	T
Myogenic factor 3	MYF3	G
Myogenic factor 4	MYF4	G
Myogenic factor 5	MYF5	G
Myoglobin		T
Myomesin 1	MYOM1	S
Myomesin 2	MYOM2	S
Myopia 1	MYP1	T
Myopia 2	MYP2	T
Myosin 15	MYO15	S
Myosin 5A	MYO5A	S
Myosin 6	MYO6	S
Myosin 7A	MYO7A	S
Myosin, cardiac	MYH7	S
Myosin, light chain 2	MYL2	S
Myosin, light chain 3	MYL3	S
Myotubularin	MTM1	S
Na <sup>+</sup> , K <sup>+</sup> ATPase, alpha	ATP1A1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 1	ATP1B1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 2	ATP1B2	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 3	ATP1B3	G
Na <sup>+</sup> /H <sup>+</sup> exchanger 1	NHE1	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 2	NHE2	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 3	NHE3	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 4	NHE4	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 5	NHE5	T
N-acetylgalactosamine-6-sulfate sulfatase	GALNS	E
N-acetylglucosamine-6-sulfatase	GNS	E
N-acetylglucosaminidase, alpha	NAGLU	E
NADH dehydrogenase		E
NADH-cytochrome b5 reductase	DIA1	E
NADPH-dependent cytochrome P450 reductase	POR	E
NB6		I
Nebulin	NEB	S
Nephrosis 1	NPHS1	T
Neural retina-specific gene	NRL	G
Neuraminidase sialidase	NEU	T
Neuregulin	HGL	G

Neurexin		N
Neuroendocrine convertase 1	NEC1, PCSK1	E
Neurokinin A	NKNA	N
Neurokinin B	NKNB	N
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Neurotensin	NTS	N
Neurotensin receptor	NTSR1	N
Nibrin	NBS1	G
Noggin	NOG	G
Notch ligand - jagged 1	JAG1, AGS	G
Nuclear factor I-kappa-B-like gene	IKBL	I
Nuclear factor kappa beta	NFKB	I
Nuclear factor of activated T cells (NFAT) complex, cytosolic	NFATC	G
Nuclear factor of activated T cells (NFAT) complex, preexisting component	NFATP	G
Ocular albinism 1	OA1	S
Oculocutaneous albinism II	OCA2	S
Oncogene ERG (early reponse gene)		G
Oncogene fos	FOS	G
Oncogene GLI1	GLI	G
Oncogene GLI2	GLI2	G
Oncogene GLI3	GLI3	G
Oncogene sis	PDGFB	G
Oncogene src		G
Opioid receptor, delta	OPRD1	N
Opioid receptor, kappa	OPRK1	N
Opioid receptor, mu	OPRM1	N
Ornithine delta-aminotransferase	OAT	E
Osteocalcin		S
Osteonectin	ON	G
Osteopontin	OPN	G
Osteoprotegerin	OPG	G
Oxytocin	OXT	N
Oxytocin receptor	OXTR	N
p21-activated kinase 3	PAK3	G
Paired box homeotic gene 1	PAX1	G
Paired box homeotic gene 2	PAX2	G
Paired box homeotic gene 3	PAX3	G
Paired box homeotic gene 6	PAX6	G
Paired box homeotic gene 7	PAX7	G
Paired box homeotic gene 8	PAX8	G
Paired-like homeodomain transcription factor 2	PITX2	G
Paired-like homeodomain transcription factor 3	PITX3	G



Parathyroid hormone	PTH	G
Parathyroid hormone receptor	PTHrP	G
Parathyroid hormone related-peptide	PTHrP	G
Parathyroid hormone-like hormone	PTHrP	G
Patched (Drosophila) homolog, PTCH	PTCH	G
Peanut-like 1	PNUTL1	I
Peripherin, PRPH		S
Peroxisomal membrane protein 1	PXMP1	S
Peroxisomal membrane protein 3	PXMP3	T
Peroxisome biogenesis factor 1	PEX1	T
Peroxisome biogenesis factor 19	PEX19	T
Peroxisome biogenesis factor 6	PEX6	T
Peroxisome biogenesis factor 7	PEX7	T
Peroxisome receptor 1	PXR1	T
Phenylethanolamine N-methyltransferase, PNMT	PNMT	E
Phosphate regulating gene with homologies to endopeptidases on the X chromosome	PHEX	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 1	PDNP1	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 2	PDNP2	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 3	PDNP3	G
Phosphofructokinase, muscle	PFKM	E
Phosphoglucose isomerase	GPI	E
Phosphoglycerate kinase 1	PGK1	E
Phosphoglycerate mutase 2	PGAM2	E
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phosphomannomutase 2	PMM2	G
Phosphoribosyl pyrophosphate synthetase	PRPS1	E
Phosphorylase kinase, alpha 1 (muscle)	PHKA1	E
Phosphorylase kinase, beta	PHKB	E
Phosphorylase kinase, delta		E
Phosphorylase kinase, gamma 2	PHKG2	E
Phytanoyl-CoA hydroxylase	PHYH	G
Pineolytic beta-receptors		E
Plakophilin 1	PKP1	T
Plasminogen	PLG	E
Platelet derived growth factor	PDGF	G
Platelet derived growth factor receptor	PDGFR	G

Plectin 1	PLEC1	T
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium voltage-gated channel E1	KCNE1	N
Potassium voltage-gated channel Q1	KCNQ1	N
Potassium voltage-gated channel Q2	KCNQ2	N
Potassium voltage-gated channel Q3	KCNQ3	N
POU domain, class 3, transcription factor 4	POU3F4	G
POU domain, class 4, transcription factor 3	POU4F3	G
Prion protein	PRNP	N
Procollagen N-protease		E
Procollagen peptidase		E
Prodynorphin		N
Profibrinolysin		G
Progesterone receptor (RU486 binding receptor)	PGR	G
Prolactin receptor	PRLR	G
Prolactin releasing hormone	PRH	G
Proliferin	PLF	G
Proopiomelanocortin	POMC	N
Properdin P factor, complement	PFC, PFD	I
Prophet of Pit1	PROP1	G
Propionyl-CoA carboxylase, alpha	PCCA	E
Prosaposin	PSAP	N
Prostacyclin synthase		I
Prostaglandin 15-OH dehydrogenase	HGPD; PGDH	I
Prostaglandin D - DP receptor		I
Prostaglandin E1 receptor		I
Prostaglandin E2 receptor		I
Prostaglandin E3 receptor		I
Prostaglandin F - FP receptor		I
Prostaglandin F2 alpha receptor		I
Prostaglandin I2 receptor		T
Prostaglandin IP receptor		I
Prostaglandin isomerase		G
Protease nexin 2	PN2	E
Protective protein for beta-galactosidase	PPGB	E
Protein C	PROC	I
Proteinase 3		I
Purine nucleoside phosphorylase	NP	E
Purinergic receptor P1A1		N
Purinergic receptor P1A2		N
Purinergic receptor P1A3		N
Purinergic receptor P2X, 1	P2RX1	N
Purinergic receptor P2X, 2	P2RX2	N
Purinergic receptor P2X, 3	P2RX3	N
Purinergic receptor P2X, 4	P2RX4	N
Purinergic receptor P2X, 5	P2RX5	N
Purinergic receptor P2X, 6	P2RX6	N

Purinergic receptor P2X, 7	P2RX7	N
Purinergic receptor P2Y, 1	P2RY1	N
Purinergic receptor P2Y, 11	P2RY11	N
Purinergic receptor P2Y, 2	P2RY2	N
Pyrroline-5-carboxylate synthetase	PYCS	E
Pyruvate kinase	PKLR	E
Rabphilin		N
Radixin	RDX	S
RAS-associated protein, RAB3A	RAB3A	N
Rathke pouch homeobox, RPX	RPX	G
Receptor tyrosine kinase (RTK), Nsk2	NSK2	G
Retinal pigment epithelium specific protein (65kD)	RPE65	S
Retinitis pigmentosa gene 1	RP1	S
Retinitis pigmentosa gene 2	RP2	S
Retinitis pigmentosa gene 3	RP3	S
Retinitis pigmentosa gene 6	RP6	S
Retinitis pigmentosa gene 7	RP7, RDS	S
Retinoblastoma 1	RB1	G
Retinoic acid receptor, alpha	RARA	G
Retinoic acid receptor, beta	RARB	G
Retinoic acid receptor, gamma	RARG	G
Retinoid X receptor, alpha	RXRA	G
Retinoid X receptor, beta	RXRB	G
Retinoid X receptor, gamma	RXRG	G
Retinol binding protein 4	RBP4	T
Rhodopsin	RHO	S
RIGUI	RIGUI	G
Rim		N
Rod outer segment membrane protein 1	ROM1	S
Ryanodine receptor 1, skeletal	RYR1	G
Serotonin N-acetyltransferase	SNAT	E
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Sex hormone binding globulin, SHBG		T
Sialoprotein, bone	BSP	G

Signal transducer and activator of transcription 1	STAT1	G
Signaling lymphocyte activation molecule	SLAM	I
Sine oculis homeobox, drosophila, homolog 1	SIX1	G
Sine oculis homeobox, drosophila, homolog 2	SIX2	G
Sine oculis homeobox, drosophila, homolog 5	SIX5	G
Sjogren (Sjogren) syndrome antigen A1	SSA1	I
Slug protein		G
Small nuclear ribonucleoprotein polypeptide N	SNRPN	S
Smoothelin	SMTN	G
Smoothened (Drosophila) homolog	SMOH	G
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage gated, type IV, alpha polypeptide	SCN4A	N
Sodium channel, voltage gated, type V, alpha polypeptide	SCN5A	N
Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Solute carrier family 1 (glutamate transporter), member 1	SLC1A1	T
Solute carrier family 1 (glutamate transporter), member 2	SLC1A2	T
Solute carrier family 12, member 1	SLC12A1	T
Solute carrier family 12, member 2	SLC12A2	T
Solute carrier family 12, member 3	SLC12A3	T
Solute carrier family 16 (monocarboxylate transporter), member 1	SLC16A1	T
Solute carrier family 16 (monocarboxylate transporter), member 7	SLC16A7	T
Solute carrier family 17, member 1	SLC17A1	T
Solute carrier family 17, member 2	SLC17A2	T
Solute carrier family 19 (folate transporter), member 1	SLC19A1	T
Solute carrier family 21, member 2	SLC21A2	T
Solute carrier family 21, member 3	SLC21A3	T
Solute carrier family 25, member 12	SLC25A12	T
Solute carrier family 6 (GAMMA-AMINOBTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Solute carrier family 6, member 10	SLC6A10	T

Solute carrier family 6, member 8	SLC6A8	T
Solute carrier family 7(amino acid transporter), member 1	SLC7A1	T
Solute carrier family 7(amino acid transporter), member 2	SLC7A2	T
Solute carrier family 7(amino acid transporter), member 7	SLC7A7	T
Solute carrier family 8 (sodium/calcium exchanger), member 1	SLC8A1	T
Somatostatin	SST	N
Somatostatin receptor, SSTR1	SSTR1	N
Somatostatin receptor, SSTR2	SSTR2	G
Somatostatin receptor, SSTR3	SSTR3	N
Somatostatin receptor, SSTR4	SSTR4	N
Somatostatin receptor, SSTR5	SSTR5	N
Sonic hedgehog, SHH	SHH	G
Sorbitol dehydrogenase	SORD	E
Sorcin	SRI	T
Spectrin alpha	SPTA1	S
Spectrin beta	SPTB	S
Sperm adhesion molecule	SPAM1	G
Sperm protamine P1	PRM1	G
Sperm protamine P2	PRM2	G
Sphingomyelinase	SMPD1	E
Split hand/foot malformation gene	DSS1	G
SRY-box 10	SOX10	G
SRY-box 11	SOX11	G
SRY-box 3	SOX3	G
SRY-box 4	SOX4	G
SRY-box 9	SOX9	G
Steroid 5 alpha reductase 1	SRD5A1	E
Steroid 5 alpha reductase 2	SRD5A2	E
Steroid sulphatase	STS	E
Substance P		N
Succinate dehydrogenase 1	SDH1	E
Succinate dehydrogenase 2	SDH2	E
Sulfamidase	SGSH	G
Superoxide dismutase 1	SOD1	E
Superoxide dismutase 3	SOD3	E
Survival of motor neuron 1, telomeric	SMN1	T
Synapsin 1a & 1b	SYN1	N
Synapsin 2a & 2b	SYN2	N
Synaptic vesicle protein 2	SV2	N
Synaptobrevin 1	SYB1	N
Synaptobrevin 2	SYB2	N
Synaptogyrin		N
Synaptophysin	SYP	N
Synaptosomal-associated protein, 25KD	SNAP25	N

Synaptotagmin 1	SYT1	N
Synaptotagmin 2	SYT2	N
Synovial sarcoma gene 1	SSX1	G
Synovial sarcoma gene 2	SSX2	G
Syntaxin 1	STX1	N
Tachykinin receptor, NK1R	TACR1	N
Tachykinin receptor, NK2R	TACR2	N
Tachykinin receptor, NK3R	TACR3	N
Talin, TLN		S
T-BOX 1	TBX1	G
T-BOX 2	TBX2	G
T-BOX 3	TBX3	G
T-BOX 4	TBX4	G
T-BOX 5	TBX5	G
T-BOX 6	TBX6	G
TEK, tyrosine kinase, endothelial	TEK	E
Telomerase protein component		E
Tetranectin	TNA	T
Thrombospondin	THBS1	G
Thromboxane A synthase 1	TBXAS1	I
Thromboxane A2	TXA2	I
Thromboxane A2 receptor	TBXA2R	I
Thymosin		I
Thyrotropin releasing hormone	TRH	N
Thyrotropin releasing hormone	TRH	G
Thyrotropin releasing hormone receptor	TRHR	N
Tip-associated protein	TAP	I
Tissue non-specific alkaline phosphatase		E
TNSAP		
Titin	TTN	S
Tocopherol (alpha) transfer protein	TTPA	T
Torticollis, keloids, cryptorchidism and renal dysplasia gene	TKCR	G
Transforming growth factor, alpha	TGFA	G
Transforming growth factor, beta 2	TGFB2	G
Transforming growth factor, beta induced	TGFBI	G
Transforming growth factor, beta receptor 2	TGFBR2	G
Transglutaminase 1	TGM1	G
Transglutaminase 2	TGM2	G
Transglutaminase 4	TGM4	G
Transthyretin	TTR	T
Treacle gene	TCOF1	G
Triosephosphate isomerase	TPI1	E
Tropomyosin 1 alpha	TPM1	S
Tropomyosin 3 (non-muscle)	TPM3	S
Troponin C		S
Troponin I	TNNI3	S
Troponin T2, cardiac	TNNT2	S

Trypsinogen 1	TRY1	E
Trypsinogen 2	TRY2	E
Tubby-like protein 1	TULP1	G
Tuberous sclerosis 1	TSC1	G
Tuberous sclerosis 2	TSC2	G
Tumor susceptibility gene 101	TSG101	G
Tumour necrosis factor (TNF) receptor associated factor 1	TRAF1	I
Tumour necrosis factor (TNF) receptor associated factor 2	TRAF2	I
Tumour necrosis factor (TNF) receptor associated factor 3	TRAF3	I
Tumour necrosis factor (TNF) receptor associated factor 4	TRAF4	I
Tumour necrosis factor (TNF) receptor associated factor 5	TRAF5	I
Tumour necrosis factor (TNF) receptor associated factor 6	TRAF6	I
Tumour necrosis factor alpha	TNFA	I
Tumour necrosis factor alpha receptor	TNFAR	I
Tumour necrosis factor beta	TNFB	I
Tumour necrosis factor beta receptor	TNFBR	I
Tumour protein p53	TP53, P53	G
Tumour protein p63	TP63	G
Tumour protein p73	TP73	G
Tumour protein, translationally-controlled 1	TPT1	G
Tumour suppressor gene DRA	DRA	I
Tyrosinase	TYR	E
Tyrosinase-related protein 1	TYRP1	E
Tyrosine aminotransferase	TAT	E
Ubiquitin activating enzyme, E1		E
Ubiquitin protein ligase E3A	UBE3A	E
Uncoupling protein 3	UCP3	T
Undulin 1	COL14A1	S
Uroporphyrinogen decarboxylase	UROD	E
Usher syndrome 2A	USH2A	S
Vacuolar proton pump, subunit 1	VPP1	N
Vacuolar proton pump, subunit 3	VPP3	N
Vascular endothelial growth factor	VEGF	G
Vasoactive intestinal polypeptide	VIP	N
Vasoactive intestinal polypeptide receptor	VIPR	N
Villin		S
Vinculin		S
Vitamin D receptor	VDR	G
Vitelliform macular dystrophy, atypical gene	VMD1	T
Von Hippel-Lindau gene	VHL	G
Von Willebrand factor	VWF	T
Werner syndrome helicase	WRN	G

Winged helix nude	WHN	G
Wingless family, wnt2	WNT2	G
Wingless family, wnt4	WNT4	G
Wingless family, wnt5	WNT5	G
Wingless family, wnt7	WNT7	G
Wingless family, wnt8	WNT8	G
Wiskott-Aldrich syndrome protein	WASP, THC	I
Wnt inhibitory factor, WIF-1	WIF1	G
Wolf-Hirschhorn syndrome candidate 1 gene	WHSC1	G
Wolfram syndrome 1 gene	WFS1	S
Xeroderma pigmentosum, complementation group A	XPA	E
Xeroderma pigmentosum, complementation group B	XPB	E
Xeroderma pigmentosum, complementation group C	XPC	E
Xeroderma pigmentosum, complementation group D		E
Xeroderma pigmentosum, complementation group E		E
Xeroderma pigmentosum, complementation group F	XPF	E
Xeroderma pigmentosum, complementation group G	ERCC5	E
X-ray repair gene	XRCC9	G

421. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 420.

422. A set according to claim 420 or 421 in which a minority of said probes for listed genes are absent.

423. A set according to claim 420 or 421 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.

424. A set according to claim 420 or 421 in which a limited number of probes are replaced by probes for non-listed genes.

425. A set of probes for a core group of genes according to any of claims 420 to 424 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.



426. A set according to any of claims 420 to 425 consisting of probes for members of a sub-group of the core group.
427. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.
428. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.
429. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.
430. A set according to claim 427 or 428 in which said substrate is a semiconductor microchip.
431. A set according to any preceding claim for use in a biological assay for detection of said gene variants.
432. A set according to any preceding claim for use in the measurement of differential gene expression levels.
433. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
434. A medical device including a set according to any of claims 420 to 432 for use in an array for detection of differential gene expression levels.
435. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 420) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 420 and 422 to 432 and relating the probe hybridisation pattern to said variations.
436. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 421) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 421 to 432 and relating the probe interaction pattern to said variations.
437. Use of a set or device according to any of claims 420 to 432 for the prognosis and management of patients suffering from or at risk of experiencing the symptoms and consequences of dysfunction, damage or disease of the skin, muscle, connective tissue or bone.
438. Use of a set or device according to any of claims 420 to 432 for predicting likely therapeutic response and adverse events following therapeutic intervention.
439. Use of a set or device according to any of claims 420 to 432 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
440. Use of a set or device according to any of claims 420 to 432 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
441. Use of a set or device according to any of claims 420 to 432 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
442. Use of a set or device according to any of claims 420 to 432 for the development of new strategies of therapeutic intervention and in clinical trials.

443. Use of a set or device according to any of claims 420 to 432 for construction of and generation of algorithms for patient and healthcare management.
444. Use of a set or device according to any of claims 420 to 432 for modelling or assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations
445. Use of a set or device according to any of claims 420 to 432 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
446. Use of a set or device according to any of claims 420 to 432 for predicting optimum configuration/management of thereapeutic intervention.
447. A method according to claim 435 or 436 in which the identification of gene variants is indicative of a higher risk of developing the symptoms and consequences of dysfunction, damage or disease of the skin, muscle, connective tissue or bone for the patient or individual.
448. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop the symptoms and consequences of dysfunction, damage or disease of the skin, muscle, connective tissue or bone, which method comprises:
- i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from the symptoms and consequences of dysfunction, damage or disease of the skin, muscle, connective tissue or bone;
  - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the the symptoms and consequences of dysfunction, damage or disease of the skin, muscle, connective tissue or bone;
  - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 420 to 426;
  - iv) calculating the frequencies of these alleles in the samples from i) and ii);
  - v) comparing the frequencies of these alleles in i) and ii);
  - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing the symptoms and consequences of dysfunction, damage or disease of the skin, muscle, connective tissue or bone.
449. A method for assessing whether a given subject will be at risk of developing symptoms, which comprises comparing said subject's genotype with a model generated by the method of claim 448.
450. A method according to any of claims 435, 436, 448 and 449 wherein at least one step is computer-controlled.
451. An assay suitable for use in a method according to any of claims 435, 436, 448 and 449; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 420 to 426 in a biological sample.
452. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing the symptoms and consequences of dysfunction, damage or disease of the skin, muscle, connective tissue or bone; said kit comprising:
- i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 420 or 422 to 426 in a sample of human DNA;
  - ii) reagents for use in the detection process

- iii) readout indicating the probability of a patient or individual developing the symptoms and consequences of dysfunction, damage or disease of the skin, muscle, connective tissue or bone.
- 453. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing the symptoms and consequences of dysfunction, damage or disease of the skin, muscle, connective tissue or bone; said kit comprising:
  - i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core group of genes as defined in any of claims 421 to 426 in an expressed-protein-containing human sample;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing the symptoms and consequences of dysfunction, damage or disease of the skin, muscle, connective tissue or bone.
- 454. A set of probes according to claim 420, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.
- 455. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to endocrine and metabolic dysfunction, damage or disease; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

#### KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME  
 T TRANSPORT & STORAGE  
 S STRUCTURAL  
 I IMMUNITY  
 N NERVOUS TRANSMISSION  
 G GROWTH & DIFFERENTIATION

METABOLIC & ENDOCRINE GENE LIST	HUGO gene symbol	Protein function
17beta hydroxysteroid dehydrogenase 1	HSD17B1	E
17beta hydroxysteroid dehydrogenase 3	HSD17B3	E
17beta hydroxysteroid dehydrogenase 4	HSD17B4	E
17beta hydroxysteroid oxidoreductase		E
17-ketosteroid reductase		N
18-hydroxysteroid oxidoreductase		E
2,3-bisphosphoglycerate mutase	BPGM	E
2,4-dienoyl CoA reductase	DECR	E
3 beta hydroxysteroid dehydrogenase 2	HSD3B2	E
3-oxoacid CoA transferase	OXCT	E

5-adenosyl homocysteine hydrolase		E
6-phosphofructo-2-kinase	PFKFB1	E
6-pyruvoyltetrahydropterin synthase	PTS	E
Acetoacetyl 1-CoA-thiolase	ACAT1	E
Acetyl CoA acyltransferase	ACAA	E
Acetyl CoA carboxylase	ACC	E
Acetyl CoA carboxylase alpha	ACACA	E
Acetylcholinesterase	ACHE	E
Acid phosphatase 2, lysosomal	ACP2	E
Actin, alpha, cardiac	ACTC	S
Actin, alpha, skeletal	ACTA1	S
Actin, alpha, smooth, aortic	ACTA2	S
Activin		G
Activin A receptor, type 2B	ACVR2B	G
Activin A receptor, type 2-like kinase 1	ACVRL1	G
Acyl CoA dehydrogenase, long chain	ACADL	E
Acyl CoA dehydrogenase, medium chain	ACADM	E
Acyl CoA dehydrogenase, short chain	ACADS	E
Acyl CoA dehydrogenase, very long chain	ACADVL	E
Acyl CoA synthetase, long chain, 1	LACS1	E
Acyl CoA synthetase, long chain, 2	LACS2	E
Acyl CoA synthetase, long chain, 4	ACS4	E
Acyl malonyl condensing enzyme		E
Adenomatous polyposis coli tumour suppressor gene	APC	G
Adenosine deaminase	ADA	E
Adenosine monophosphate deaminase	AMPD	E
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N
Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenyl cyclase		N
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adenylate transferase		E
ADP-ribosyltransferase	ADPRT	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N

Adrenoleukodystrophy gene	ALD	E
Albumin, ALB	ALB	T
Alcohol dehydrogenase 1	ADH1	E
Alcohol dehydrogenase 2	ADH2	E
Alcohol dehydrogenase 3	ADH3	E
Alcohol dehydrogenase 4	ADH4	E
Alcohol dehydrogenase 5	ADH5	E
Alcohol dehydrogenase 6	ADH6	E
Alcohol dehydrogenase 7	ADH7	E
Aldehyde dehydrogenase 1	ALDH1	E
Aldehyde dehydrogenase 10	ALDH10	E
Aldehyde dehydrogenase 2	ALDH2	E
Aldehyde dehydrogenase 5	ALDH5	E
Aldehyde dehydrogenase 6	ALDH6	E
Aldehyde dehydrogenase 7	ALDH7	E
Aldolase A	ALDOA	E
Aldolase B	ALDOB	E
Aldolase C	ALDOC	E
Aldosterone receptor	MLR	G
Alkaline phosphatase, liver/bone/kidney	ALPL	T
Alkylglycerone phosphate synthase	AGPS	E
Alpha 1 acid glycoprotein	AAG; AGP	T
alpha1-antitrypsin	PI	E
alpha-actinin 2	ACTN2	G
alpha-actinin 3	ACTN3	G
alpha-amino adipic semialdehyde synthase		E
alpha-glucosidase, neutral AB	GANAB	E
alpha-glucosidase, neutral C	GANC	E
alpha-ketoglutarate dehydrogenase		E
Aminomethyltransferase	AMT	E
Aminopeptidase P	XPNPEP2	E
Amphiregulin	AREG	G
Amylo-1,6-glucosidase	AGL	E
Androgen receptor	AR	G
Angiopoietin 1	ANGPT1	G
Angiopoietin 2	ANGPT2	G
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensin receptor 1	AGTR1	T
Angiotensin receptor 2	AGTR2	T
Angiotensinogen	AGT	E
Anti-Mullerian hormone	AMH	G
Anti-Mullerian hormone type 2 receptor	AMHR2	G
Apolipoprotein A I	APOA1	T
Apolipoprotein A II	APOA2	T
Apolipoprotein B	APOB	T
Apolipoprotein C1	APOC1	T
Apolipoprotein C2	APOC2	T
Apolipoprotein C3	APOC3	T

Apolipoprotein D	APOD	T
Apolipoprotein E	APOE	T
Apolipoprotein H	APOH	T
Aquaporin 1	AQP1	T
Aquaporin 2	AQP2	T
Arginine vasopressin	AVP	N
Arginine vasopressin receptor 1A	AVPR1A	N
Arginine vasopressin receptor 1B	AVPR1B	N
Arginine vasopressin receptor 2	AVPR2	N
Asparagine synthetase	AS	E
Aspartate transcarbamoylase		E
Ataxia telangiectasia complementation group D	ATD, ATDC	G
Ataxia telangiectasia gene, AT	ATM	G
ATP cobalamin adenosyltransferase		E
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G
Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
Attractin		I
Autoimmune regulator, AIRE	AIRE	I
beta-endorphin receptor		N
beta-galactosidase	GLB1	E
beta-ketoacyl reductase		E
Bile acid coenzyme A: amino acid N-acyltransferase	BAAT	E
Bile salt export pump	BSEP, PFIC2	T
Bile salt-stimulated lipase	CEL	E
Bilirubin UDP-glucuronosyltransferase		E
Bloom syndrome protein	BLM	G
Bradykinin receptor B1		I
Bradykinin receptor B2		I
Branched chain aminotransferase 1, cytosolic	BCAT1	E
Branched chain aminotransferase 2, mitochondrial	BCAT2	E
Branched chain keto acid dehydrogenase E1, BCKDHA alpha polypeptide		E
Branched chain keto acid dehydrogenase E1, BCKDHB beta polypeptide		E
Butyrylcholinesterase	BCHE	E
C17-20 desmolase		E
C3 convertase		E
Calbindin 1	CALB1	G
Calbindin D9K	CALB3	G
Calcineurin A1	CALNA1	I
Calcineurin A2	CALNA2	I
Calcineurin A3	CALNA3	I
Calcineurin B		I

Calcitonin receptor /Calcitonin gene-related peptide receptor	CALCR	N
Calcitonin/Calcitonin gene-related peptide alpha	CALCA	N
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha-1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha-1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha-1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha-1E (CACNL1A6)	CACNA1E	N
Calcium channel, voltage-dependent, Alpha-2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, L type, alpha 1S subunit	CACNA1S	N
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N
Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	CACNA1A	N
Calcium channel, voltage-dependent, T-type		N
Calcium sensing receptor	CASR	T
Calmodulin 1	CALM1	G
Calmodulin 2	CALM2	G
Calmodulin 3	CALM3	G
Calmodulin-dependant protein kinase II	CAMK2A	G
Calnexin	CANX	G
Calpain	CAPN, CAPN3	E
Calretinin	CALB2	N
Canalicular multispecific organic anion transporter	CMOAT	T
Cannabinoid receptor	CNR1	N
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Carboxylesterase 1	CES1	E
Cardiac-specific homeobox, CSX	CSX	G
Carnitine acetyltransferase	CRAT	E
Carnitine acylcarnitine translocase	CACT	E
Carnitine palmitoyltransferase I	CPT1A	E
Carnitine palmitoyltransferase II	CPT2	E
Carnitine transporter protein	CDSP, SCD	T
Carnosinase		N

Cartilage-hair hypoplasia gene	CHH	N
Catechol-O-methyltransferase	COMT	E
Cell adhesion molecule, intercellular, ICAM	ICAM1	G
Cell adhesion molecule, leukocyte- endothelial, LECAM (CD62)	LECAM1	G
Cell adhesion molecule, liver, LCAM	LCAM	G
Cell adhesion molecule, neural, NCAM1	NCAM1	G
Cell adhesion molecule, neural, NCAM120	NCAM120	G
Cell adhesion molecule, neural, NCAM2	NCAM2	G
Cell adhesion molecule, platelet-endothelial, PECAM	PECAM1	G
Cell adhesion molecule, vascular, VCAM	VCAM1	G
c-erbB2	ERBB2	G
c-erbB3	ERBB3	G
c-erbB4	ERBB4	G
Chitotriosidase	chit	E
Cholecystokinin	CCK	N
Cholecystokinin B receptor	CCKBR	N
Cholesterol ester hydroxylase		E
Cholesterol ester transfer protein	CETP	T
Choline acetyltransferase	CHAT	E
Chromogranin A	CHGA	G
Chymase	CHY1	
Citrate synthase		E
Clathrin		T
Clusterin	CLU	G
CoA transferase		E
Collagen IV alpha 5	COL4A5	S
Collagen IV alpha 6	COL4A6	S
Complex III		E
Complex V	MTATP6	E
Corticosteroid binding globulin	CBG	N
Corticotrophin-releasing hormone	CRH	T
Corticotrophin-releasing hormone receptor	CRHR1	T
Cortisol receptor		I
Cubilin	CUBN	T
Cyclic AMP-dependent protein kinase	PKA	E
Cyclic nucleotide phosphodiesterase 1B	PDE1B	E
Cyclic nucleotide phosphodiesterase 1B1	PDE1B1	E
Cyclic nucleotide phosphodiesterase 2A3	PDE2A3	E
Cyclic nucleotide phosphodiesterase 3A	PDE3A	E
Cyclic nucleotide phosphodiesterase 3B	PDE3B	E
Cyclic nucleotide phosphodiesterase 4A	PDE4A	E
Cyclic nucleotide phosphodiesterase 4C	PDE4C	E
Cyclic nucleotide phosphodiesterase 5A	PDE5A	E
Cyclic nucleotide phosphodiesterase 6A	PDE6A	E
Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E



Cyclic nucleotide phosphodiesterase 8	PDE8	E
Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclin-dependent kinase inhibitor 1C (P57, KIP2)	CDKN1C	G
Cyclin-dependent kinase inhibitor 2A (p16)	CDKN2A	G
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
CYP11A1	CYP11A1	E
CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E
CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E
CYP2C18	CYP2C18	E
CYP2C19	CYP2C19	E
CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E
CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E
CYP2F1	CYP2F1	E
CYP2J2	CYP2J2	E
CYP3A3	CYP3A3	E
CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystathionase	CTH	E
Cystathione beta synthase	CBS	E
Cystic fibrosis transmembrane conductance	CFTR	N

regulator, CFTR		
Cystinosis	CTNS	T
Cytidine deaminase	CDA	E
Cytidine-5-prime-triphosphate synthetase	CTPS	E
Cytochrome a		E
Cytochrome c		E
Cytochrome c oxidase, MTCO		E
Cytokine-suppressive antiinflammatory drug-binding protein 1	CSBP1	I
Cytokine-suppressive antiinflammatory drug-binding protein 2	CSBP2	I
DAX1 nuclear receptor	DAX1	I
D-beta-hydroxybutyrate dehydrogenase		E
Dehydratase		E
Delta 4-5 oxosteroid isomerase		E
Delta aminolevulinate synthase 1	ALAS1	E
Delta aminolevulinate synthase 2	ALAS2	E
Deoxycorticosterone (DOC) receptor		E
Deoxyuridine triphosphatase; dUTPase		E
DHEA sulfotransferase	STD	E
Dihydrodiol dehydrogenase 1	DDH1	E
Dihydrolipoamide branched chain transacylase	DBT	N
Dihydrolipoamide dehydrogenase	DLD	N
Dihydrolipoyl dehydrogenase 2	PDHA	E
Dihydrolipoyl transacetylase	PDHA	E
Dihydroorotase		E
Dihydropyrimidinase	DPYS	E
Dihydroxyacetonephosphate acyltransferase	DHAPAT	E
Dihydropyrimidine dehydrogenase	DPYD	E
DNA glycosylases		E
DNA helicases		E
DNA Ligase 1	LIG1	E
DNA methyltransferase	DNMT	E
DOPA decarboxylase	DDC	E
Dopamine beta hydroxylase	DBH	E
Dopamine receptors D1	DRD1	N
Dopamine receptors D2	DRD2	N
Dopamine receptors D3	DRD3	N
Dopamine receptors D4	DRD4	N
Dopamine receptors D5	DRD5	N
Dynamin	DNM1	G
Electron-transferring-flavoprotein alpha	ETFA	T
Electron-transferring-flavoprotein beta	ETFB	T
Electron-transferring flavoprotein dehydrogenase	ETFDH	E
Endometrial bleeding-associated factor	EBAF	G
Endothelin converting enzyme	ECE1	N

Endothelin receptor type A	EDNRA	N
Endothelin receptor type B	EDNRB	N
Enolase	ENO1	E
Enoyl CoA reductase		E
Enterokinase	PRSS7, ENTK	E
Ephrin receptor tyrosine kinase A	EPHA	G
Ephrin receptor tyrosine kinase B	EPHB	G
Epidermal growth factor	EGF	G
Epidermal growth factor receptor	EGFR	G
Erythropoietin	EPO	I
Estrogen receptor	ESR	G
Excision repair complementation group 1 protein	ERCC1	E
Factor 1 (No. one)	F1	I
FADH dehydrogenase		E
Fatty acid binding proteins FABP2	FABP2	T
Fc fragment of IgG, high affinity IA, receptor for	FCGR1A	G
Fc fragment of IgG, low affinity IIa, receptor for (CD32)	FCGR2A	G
Fc fragment of IgG, low affinity IIIa, receptor for (CD16)	FCGR3A	G
Ferritin, H subunit		T
Ferritin, L subunit	FTL	T
Fibrinogen alpha	FGA	S
Fibrinogen beta	FGB	S
Fibrinogen gamma	FGG	S
Fibroblast growth factor	FGF1	G
Fibroblast growth factor receptor 1	FGFR1	G
Fibroblast growth factor receptor 2	FGFR2	G
Fibroblast growth factor receptor 3	FGFR3	G
Flavin-containing monooxygenase 1	FMO1	E
Flavin-containing monooxygenase 2	FMO2	E
Flavin-containing monooxygenase 3	FMO3	E
Flavin-containing monooxygenase 4	FMO4	E
Follicle stimulating hormone receptor	FSHR, ODG1	G
Follicle stimulating hormone, FSH	FSHB	G
Follistatin		G
Frataxin	FRDA	G
Fructose-1,6-diphosphatase	FBP1	E
Fumarase	FH	E
Fumarylacetoacetase	FAH	E
GABA receptor, alpha 1	GABRA1	N
GABA receptor, alpha 2	GABRA2	N
GABA receptor, alpha 3	GABRA3	N
GABA receptor, alpha 4	GABRA4	N
GABA receptor, alpha 5	GABRA5	N
GABA receptor, alpha 6	GABRA6	N

GABA receptor, beta 1	GABRB1	N
GABA receptor, beta 2	GABRB2	N
GABA receptor, beta 3	GABRB3	N
GABA receptor, gamma 1	GABRG1	N
GABA receptor, gamma 2	GABRG2	N
GABA receptor, gamma 3	GABRG3	N
GABA transaminase	ABAT	E
Galactocerebrosidase	GALC	E
Galactokinase	GALK1	E
Galactose 1-phosphate uridyl-transferase	GALT	E
Galanin	GAL	N
Galanin receptor	GALNR1	N
Gamma-glutamyl carboxylase	GGCX	T
Gamma-glutamyltransferase 1	GGT1	T
Gamma-glutamyltransferase 2	GGT2	T
Gap junction protein beta 1	GJB1	T
Gap junction protein beta 3	GJB3	T
Gastric inhibitory polypeptide GIP	GIP	T
Gastric inhibitory polypeptide receptor, GIPR	GIPR	T
Gastric Intrinsic factor, GIF	GIF	E
Gastric lipase, LIPF		T
Gastrin	GAS	G
Gastrin releasing peptide	GRP	T
Gastrin releasing peptide receptor	GRPR	T
Glucagon receptor	GCGR	G
Glucagon synthase		T
Glucagon-like peptide receptor 1	GLP1R	G
Glucocorticoid receptor	GRL	G
Glucokinase	GCK	E
Glucosaminyl (N-acetyl) transferase 2, 1- branching enzyme	GCNT2	E
Glucose-6-phosphatase	G6PC	E
Glucose-6-phosphatase translocase	G6PT1	E
Glucose-6-phosphate dehydrogenase	G6PD	E
Glucosidase, acid beta	GBA	E
Glutamate decarboxylase, GAD	GAD1	E
Glutamate dehydrogenase	GLUD1	E
Glutamine phosphoribosylpyrophosphate amidotransferase/PRPP amidotransferase		E
Glutamine synthase		E
Glutathione	GSH	T
Glutathione peroxidase, GPX2	GPX2	E
Glutathione reductase, GSR	GSR	E
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glutathione synthetase	GSS	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH		E
GAPDH		
Glycerol kinase	GK	E

Glycerophosphate dehydrogenase 2	GPD2	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
Glycine dehydrogenase	GLDC	E
Glycogen branching enzyme	GBE1	E
Glycogen phosphorylase	PYGL	E
Glycogen synthase 1 (muscle)	GLYS1	E
Glycogen synthase 2 (liver)	GYS2	E
Glycosyltransferases, ABO blood group	ABO	E
Gonadotropin releasing hormone	GNRH	G
Gonadotropin releasing hormone receptor	GNRHR	G
Growth arrest-specific homeobox	GAX	G
Growth hormone 1	GH1	G
Growth hormone 2 (placental)	GH2	G
Growth hormone receptor	GHR	G
Growth hormone releasing hormone (GHRH)	GHRH	G
Growth hormone releasing hormone receptor	GHRHR	G
GTP cylcohydrolase 1	GCH1	G
GTPase-activating protein, GAP	RASA1	G
Guanidinoacetate N-methyltransferase	GAMT	E
Guanine nucleotide-binding protein, alpha activating activity polypeptide, GNAO	GNAO1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 1, GNAI1	GNAI1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 2, GNAI2	GNAI2	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 3, GNAI3	GNAI3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS1	GNAS1	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS2	GNAS2	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS3	GNAS3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS4	GNAS4	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT1	GNAT1	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT2	GNAT2	N
Guanine nucleotide-binding protein, beta polypeptide 3	GNB3	N
Guanine nucleotide-binding protein, gamma polypeptide 5	GNG5	N
Guanine nucleotide-binding protein, q polypeptide	GNAQ	N
Guanylate cyclase 2D, membrane (retina- specific)	GUCY2D	E

Guanylate cyclase activator 1A (retina)	GUCA1A	E
Guanylate kinase		E
Guanylin	GUCA2	T
Guanylyl cyclase		E
Heat shock protein, HSP60		I
Heat shock protein, HSP70		I
Heat shock protein, HSP90		I
Heat shock protein, HSPA1		I
Heat shock protein, HSPA2		I
Hemopexin	HPX	I
Heparin binding epidermal growth factor	HBEGF	G
Hepatic lipase	LIPC	E
Hepatic nuclear factor-3-beta	HNF3B	E
Hepatic nuclear factor-4-alpha	HNF4A	E
Hexokinase 1	HK1	E
Hexokinase 2	HK2	E
Hexosaminidase A	HEXA,TSD	E
Hexosaminidase B	HEXB	E
Histamine receptors, H1		N
Histamine receptors, H2		N
Histamine receptors, H3		N
HMG-CoA lyase	HMGCL	E
HMG-CoA reductase	HMGCR	E
HMG-CoA synthase	HMGCS2	E
Holocarboxylase synthetase	HLCS	E
Holoprosencephaly 1	HPE1	G
Holoprosencephaly 2	HPE2	G
Holoprosencephaly 3	HPE3	G
Holoprosencephaly 4	HPE4	G
Homeobox (HOX) gene A13	HOXA13	G
Hormone-sensitive lipase	HSL	E
HSSB, replication protein		E
Human chorionic gonadotrophin, hCG	CG	G
Human placental lactogen	CSH1	G
Hydroxyacyl glutathione hydrolase	HAGH	E
Hypoxanthine-guanine phosphoribosyltransferase, HGPRT	HPRT	E
Hypoxia inducible factor 1	HIF1A	E
Hypoxia inducible factor 2		E
Iduronate 2 sulphatase	IDS	E
Immunoglobulin E (IgE) responsiveness gene	IGER	I
Immunoglobulin E (IgE) serum concentration regulator gene	IGES	I
Immunoglobulin gamma (IgG) 2	IGHG2	I
Indian hedgehog, ihh	IHH	G
Inhibin, alpha	INHA	G
Inhibin, beta A	INHBA	G
Inhibin, beta B	INHBB	G

Inhibin, beta C	INHBC	G
Inosine monophosphate dehydrogenase, IMPDH		E
Inosine triphosphatase	ITPA	E
Inositol 1,4,5-triphosphate receptor 1	ITPR1	G
Inositol monophosphatase	IMPA1	N
Inositol polyphosphate 1-phosphatase	INPP1	N
Insulin	INS	G
Insulin receptor	INSR	G
Insulin receptor substrate-1	IRS1	G
Insulin-like growth factor 1	IGF1	G
Insulin-like growth factor 1 receptor	IGF1R	G
Insulin-like growth factor 2	IGF2	G
Insulin-like growth factor 2 receptor	IGF2R	G
Integrin beta 1	ITGB1	G
Integrin beta 2	ITGB2	G
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) 10	IL10	I
Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I
Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
Iodothyronine-5'-deiodinase, type 1 and 2		E
IP3 kinase		E
Islet amyloid polypeptide	IAPP	N

Isocitrate dehydrogenase		E
Isovaleric acid CoA dehydrogenase	IVD	E
Janus kinase 1	JAK1	G
Janus kinase 2	JAK2	G
Janus kinase 3	JAK3	G
Kallman syndrome gene 1	KAL1	G
Ketohexokinase	KHK	E
ketolase		E
Lactase		E
Lactotransferrin	LTF	T
Laminin 5, alpha 3	LAMA3	G
Laminin 5, beta 3	LAMB3	G
Laminin receptor 1	LAMR1	G
Lecithin-cholesterol acyltransferase	LCAT	E
Leptin	LEP	G
Leptin receptor	LEPR	G
Leukotriene C4 synthase	LTC4S	E
LH/choriogonadotropin (CG) receptor	LHCGR	G
Lipoamide dehydrogenase	OGDH	E
Lipoprotein lipase	LPL	I
Lipoprotein, High Density	HDLDT1	T
Lipoprotein, Intermediate Density		T
Lipoprotein, Low Density 1		T
Lipoprotein, Low Density 2		T
Lipoprotein, Very Low Density	VLDLR	T
Lipoprotein-associated coagulation factor	LACI	I
Lipoxygenase		E
Lipoxygenase 12 (platelets)	LOG12	I
Lipoxygenase 5 (leukocytes)		I
Luteinizing hormone, beta chain	LHB	G
Lymphocyte-specific protein tyrosine kinase	LCK	I
Lysosomal acid lipase	LIPA	E
MAD (mothers against decapentaplegic, Drosophila) homologue 2	MADH2	G
Malate dehydrogenase, mitochondrial	MDH2	E
Malonyl CoA decarboxylase		E
Malonyl CoA transferase		E
Maltase-glucoamylase		E
Mannosidase, alpha B lysosomal	MANB	E
Mannosyl (alpha-1,6-)-glycoprotein beta-1, 2-	MGAT2	T
N-acetylglucosaminyltransferase		
Marenostrin	MEFV	T
Matrix Gla protein	MGP	G
MEK kinase, MEKK		E
Melanocortin 2 receptor	MC2R	T
Melanocortin 4 receptor	MC4R	T
Menin	MEN1	G
Methionine adenosyltransferase	MAT1A, MAT2A	E



Methionine synthase	MTR	E
Methionine synthase reductase	MTRR	E
Methylguanine-DNA methyltransferase	MGMT	E
Methylmalonyl-CoA mutase	MUT	E
Mitochondrial trifunctional protein, alpha subunit	HADHA	E
Mitochondrial trifunctional protein, beta subunit	HADHB	E
Molybdenum cofactor synthesis 1	MOCS1	E
Molybdenum cofactor synthesis 2	MOCS2	E
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Multidrug resistance associated protein	MRP	G
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Muscle phosphorylase	PYGM	E
Na <sup>+</sup> , K <sup>+</sup> ATPase, alpha	ATP1A1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 1	ATP1B1	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 2	ATP1B2	G
Na <sup>+</sup> , K <sup>+</sup> ATPase, beta 3	ATP1B3	G
Na <sup>+</sup> /H <sup>+</sup> exchanger 1	NHE1	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 2	NHE2	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 3	NHE3	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 4	NHE4	T
Na <sup>+</sup> /H <sup>+</sup> exchanger 5	NHE5	T
N-acetyltransferase 1	NAT1	E
N-acetyltransferase 2	NAT2	E
NADH dehydrogenase (ubiquinone) Fe-S protein 1	NDUFS1	E
NADH dehydrogenase (ubiquinone) Fe-S protein 4	NDUFS4	E
NADH dehydrogenase (ubiquinone) flavoprotein 1	NDUFV1	E
NADH-cytochrome b5 reductase	DIA1	E
NADPH-dependent cytochrome P450 reductase	POR	E
Nephronophthisis 1	NPHP1	T
Nephrosis 1	NPHS1	T
Nerve growth factor	NGF	G
Nerve growth factor receptor	NGFR	G
Neuraminidase sialidase	NEU	T
Neuregulin	HGL	G
Neuroendocrine convertase 1	NEC1, PCSK1	E
Neurofibromin 1	NF1	G
Neurofibromin 2	NF2	G

Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Neurotensin	NTS	N
Neurotensin receptor	NTSR1	N
Neurotrophin 3	NTF3 or NT3	G
Neutral endopeptidase		E
Niemann-Pick disease protein	NPC1	T
Nitric oxide synthase 1, NOS1	NOS1	E
Nitric oxide synthase 2, NOS2	NOS2	E
Nitric oxide synthase 3, NOS3	NOS3	E
Notch ligand - jagged 1	JAG1, AGS	G
Nucleoside diphosphate kinase-A	NDPKA	E
Oncogene ret	RET	G
Oncogene sis	PDGFB	G
Orexin	OX	G
Orexin 1 receptor	OX1R	G
Orexin 2 receptor	OX2R	G
Ornithine delta-aminotransferase	OAT	E
Ornithine transcarbamoylase	OTC, NME1	E
Oxytocin	OXT	N
Oxytocin receptor	OXTR	N
Paired box homeotic gene 6	PAX6	G
Paired box homeotic gene 8	PAX8	G
Palmitoyl-protein thioesterase	PPT	T
Pancreatic lipase	PNLIP	E
Paraoxonase PON1	PON1	E
Paraoxonase PON2	PON2	E
Paraoxonase PON3		E
Parathyroid hormone	PTH	G
Parathyroid hormone receptor	PTHr1	G
Parathyroid hormone related-peptide	PTHrP	G
Parathyroid hormone-like hormone	PTHrLH	G
Peanut-like 1	PNUTL1	I
Peptidylglycine alpha-amidating monooxygenase	PAM	E
Peroxidase, salivary	SAPX	E
Peroxisomal membrane protein 3	PXMP3	T
Peroxisome biogenesis factor 1	PEX1	T
Peroxisome biogenesis factor 19	PEX19	T
Peroxisome biogenesis factor 6	PEX6	T
Peroxisome biogenesis factor 7	PEX7	T
Peroxisome proliferative activated receptor, alpha	PPARA	T
Peroxisome proliferative activated receptor, gamma	PPARG	T
P-glycoprotein 1	PGY1	T
P-glycoprotein 3	PGY3	T

Phenylalanine hydroxylase	PAH	E
Phenylalanine monooxygenase		E
Phenylethanolamine N-methyltransferase, PNMT	PNMT	E
Phosphodiesterase 1 / nucleotide pyrophosphatase 1	PDNP1	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 2	PDNP2	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 3	PDNP3	G
Phosphoenolpyruvate carboxykinase	PCK1	E
Phosphofructokinase, liver	PFKL	E
Phosphofructokinase, muscle	PFKM	E
Phosphoglucomutase		E
Phosphoglucose isomerase	GPI	E
Phosphoglycerate kinase 1	PGK1	E
Phosphoglycerate mutase 2	PGAM2	E
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C alpha		I
Phospholipase C beta		I
Phospholipase C delta	PLCD1	I
Phospholipase C epsilon		I
Phospholipase C gamma	PLCG1	I
Phosphomannomutase 2	PMM2	G
Phosphomannomutase-2	PMM2	T
Phosphomannose isomerase-1, PMI1	MPI	T
Phosphoribosyl pyrophosphate synthetase	PRPS1	E
Phosphorylase kinase deficiency, liver	PHK	E
Phosphorylase kinase, alpha 1 (muscle)	PHKA1	E
Phosphorylase kinase, alpha 2	PHKA2	E
Phosphorylase kinase, beta	PHKB	E
Phosphorylase kinase, delta		E
Phosphorylase kinase, gamma 2	PHKG2	E
Phytanoyl-CoA hydroxylase	PHYH	G
Pineolytic beta-receptors		E
Pituitary adenylate cyclase activating peptide	PACAP	N
Pituitary adenylate cyclase activating peptide receptor	PACAP1R	N
Plasminogen activator receptor, Urokinase	UPAR; PLAUR	S
Plasminogen activator, Tissue	PLAT; TPA	E
Plasminogen activator, Urokinase	UPA; PLAU	E

Platelet derived growth factor	PDGF	G
Platelet derived growth factor receptor	PDGFR	G
Poly (ADP-ribose) synthetase	PARS	E
Polycystin 1	PKD1	T
Polycystin 2	PKD2	T
Porphobilinogen deaminase	HMBS	E
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium inwardly-rectifying channel J11	KCNJ11	N
Potassium voltage-gated channel A1	KCNA1	N
Potassium voltage-gated channel E1	KCNE1	N
Potassium voltage-gated channel Q1	KCNQ1	N
Preproenkephalin	PENK	N
Preproglucagon	GCG;GLP1; GLP2	G
Preproglucagon		T
Preproinsulin		T
Profibrinolysin		G
Progesterone receptor (RU486 binding receptor)	PGR	G
Prolactin	PRL	G
Prolactin receptor	PRLR	G
Prolactin releasing hormone	PRH	G
Proliferin	PLF	G
Proline dehydrogenase	PRODH	E
Proline-rich protein BstNI subfamily 1	PRB1	S
Proline-rich protein BstNI subfamily 3	PRB3	S
Proline-rich protein BstNI subfamily 4	PRB4	S
Pro-melanin-concentrating hormone	PMCH	G
Proopiomelanocortin	POMC	N
Prophet of Pit1	PROP1	G
Prostacyclin synthase		I
Prostaglandin (PG) D synthase, hematopoietic	PGDS	E
Prostaglandin 15-OH dehydrogenase	HGPD; PGDH	I
Prostaglandin D - DP receptor		I
Prostaglandin E1 receptor		I
Prostaglandin E2 receptor		I
Prostaglandin E3 receptor		I
Prostaglandin F - FP receptor		I
Prostaglandin I2 receptor		T
Prostaglandin IP receptor		I
Prostaglandin isomerase		G
Prostasin, PRSS8	PRSS8	E
Protease nexin 2	PN2	E
Protein kinase B	PRKB	
Protein kinase C, alpha	PRKCA	E
Protein S	PROS1	I
Protoporphyrinogen oxidase	PPOX	E
Pterin-4-alpha-carbinolamine	PCBD	

Pyrroline-5-carboxylate synthetase	PYCS	E
Pyruvate carboxylase	PC	E
Pyruvate decarboxylase	PDHA	E
Pyruvate kinase	PKLR	E
Quinoid dihydropteridine reductase	QDPR	E
Rathke pouch homeobox, RPX	RPX	G
Relaxin H1	RLN1	G
Relaxin H2	RLN2	G
Renin	REN	E
Replication factor C	RFC2	E
Retinal pigment epithelium specific protein (65kD)	RPE65	S
Retinaldehyde binding protein 1	RLBP1	T
Retinoic acid receptor, alpha	RARA	G
Retinoic acid receptor, beta	RARB	G
Retinoic acid receptor, gamma	RARG	G
Retinoid X receptor, alpha	RXRA	G
Retinoid X receptor, beta	RXRB	G
Retinoid X receptor, gamma	RXRG	G
Retinol binding protein 1		T
Retinol binding protein 2		T
Ribosephosphate pyrophosphokinase		E
RIGUI	RIGUI	G
Ryanodine receptor 1, skeletal	RYR1	G
S100 calcium-binding protein A1	S100A1	N
S100 calcium-binding protein A2	S100A2	N
S100 calcium-binding protein A3	S100A3	N
S100 calcium-binding protein A4	S100A4	N
S100 calcium-binding protein A5	S100A5	N
S100 calcium-binding protein A6	S100A6	N
S100 calcium-binding protein A7	S100A7	N
S100 calcium-binding protein A8	S100A8	N
S100 calcium-binding protein A9	S100A9	N
S100 calcium-binding protein B	S100B	N
S100 calcium-binding protein P	S100P	N
S-adenosylmethionine decarboxylase, AMD		E
Salivary amylase, AMY1		T
Secretin	SCT	T
Secretin receptor, SCTR	SCTR	T
Serine hydroxymethyltransferase	SHMT	E
Serotonin N-acetyltransferase	SNAT	E
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N

Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Serum amyloid A	SAA	T
Serum amyloid P	SAP	T
Sex determining region Y, SRY	SRY	G
Sex hormone binding globulin, SHBG		T
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Solute carrier family 1 (amino acid transporter), member 6	SLC1A6	T
Solute carrier family 1 (neutral amino acid transporter), member 4	SLC1A4	T
Solute carrier family 10 (sodium/bile acid cotransporter family), member 1	SLC10A1	T
Solute carrier family 10 (sodium/bile acid cotransporter family), member 2	SLC10A2	T
Solute carrier family 12, member 1	SLC12A1	T
Solute carrier family 12, member 2	SLC12A2	T
Solute carrier family 12, member 3	SLC12A3	T
Solute carrier family 14, member 2	SLC14A2	T
Solute carrier family 15 (H <sup>+</sup> /peptide transporter, intestinal), member 1	SLC15A1	T
Solute carrier family 15 (H <sup>+</sup> /peptide transporter, kidney), member 2	SLC15A2	T
Solute carrier family 16 (monocarboxylate transporter), member 1	SLC16A1	T
Solute carrier family 16 (monocarboxylate transporter), member 7	SLC16A7	T
Solute carrier family 17, member 1	SLC17A1	T
Solute carrier family 17, member 2	SLC17A2	T
Solute carrier family 2 (facilitated glucose transporter), member 1	SLC2A1	T
Solute carrier family 2 (facilitated glucose transporter), member 2	SLC2A2	T
Solute carrier family 2 (facilitated glucose transporter), member 3	SLC2A3	T
Solute carrier family 2 (facilitated glucose transporter), member 4	SLC2A4	T
Solute carrier family 2 (facilitated glucose transporter), member 5	SLC2A5	T

transporter), member 5		
Solute carrier family 20, member 3	SLC20A3	T
Solute carrier family 21, member 2	SLC21A2	T
Solute carrier family 21, member 3	SLC21A3	T
Solute carrier family 22, member 1	SLC22A1	T
Solute carrier family 22, member 2	SLC22A2	T
Solute carrier family 22, member 5	SLC22A5	T
Solute carrier family 3 (facilitated glucose transporter), member 1	SLC3A1	T
Solute carrier family 4 (anion exchanger), member 1	SLC4A1	T
Solute carrier family 4 (anion exchanger), member 2	SLC4A2	T
Solute carrier family 4 (anion exchanger), member 3	SLC4A3	T
Solute carrier family 5 (sodium/glucose transporter), member 1	SLC5A1	T
Solute carrier family 5 (sodium/glucose transporter), member 2	SLC5A2	T
Solute carrier family 5 (sodium/glucose transporter), member 5	SLC5A5	T
Solute carrier family 5, member 3	SLC5A3	T
Solute carrier family 6 (GAMMA-AMINOBUTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Solute carrier family 6 (neurotransmitter transporter, serotonin), member 4	SLC6A4	T
Solute carrier family 6, member 10	SLC6A10	T
Solute carrier family 6, member 6	SLC6A6	T
Solute carrier family 6, member 8	SLC6A8	T
Solute carrier family 7(amino acid transporter), member 1	SLC7A1	T
Solute carrier family 7(amino acid transporter), member 2	SLC7A2	T
Solute carrier family 7(amino acid transporter), member 7	SLC7A7	T
Solute carrier family 8 (sodium/calcium exchanger), member 1	SLC8A1	T
Somatostatin	SST	N
Somatostatin receptor, SSTR1	SSTR1	N
Somatostatin receptor, SSTR2	SSTR2	G
Somatostatin receptor, SSTR3	SSTR3	N
Somatostatin receptor, SSTR4	SSTR4	N
Somatostatin receptor, SSTR5	SSTR5	N

Somatotrophin		G
Sorcin	SRI	T
SOS1 guanine nucleotide exchange factor	SOS1	G
Sperm protamine P1	PRM1	G
Sperm protamine P2	PRM2	G
Sphingomyelinase	SMPD1	E
SRY-box 10	SOX10	G
SRY-box 11	SOX11	G
SRY-box 3	SOX3	G
SRY-box 4	SOX4	G
SRY-box 9	SOX9	G
Steroid sulphotase	STS	E
Steroidogenic acute regulatory protein	STAR	T
Substance P		N
Succinyl CoA synthase		E
Sucrase		E
Sulfonylurea receptor	SUR	G
Superoxide dismutase 1	SOD1	E
Superoxide dismutase 3	SOD3	E
Surfeit 1	SURF1	G
T-BOX 1	TBX1	G
T-BOX 3	TBX3	G
Thiolase, peroxisomal		E
Thiopurine S-methyltransferase	TPMT	E
Thrombospondin	THBS1	G
Thromboxane A synthase 1	TBXAS1	I
Thromboxane A2	TXA2	I
Thromboxane A2 receptor	TBXA2R	I
Thymopoietin	TMPO	G
Thymosin		I
Thyroglobulin	TG	G
Thyroid hormone receptor, alpha	THRA	G
Thyroid hormone receptor, beta	THRB	G
Thyroid peroxidase	TPO	G
Thyroid receptor auxiliary protein	TRAP	G
Thyroid-stimulating hormone receptor	TSHR	G
Thyroid-stimulating hormone, alpha	TSHA	G
Thyroid-stimulating hormone, beta	TSHB	G
Thyrotropin releasing hormone	TRH	G
Thyrotropin releasing hormone receptor	TRHR	G
Thyroxin-binding globulin	TBG	T
Transacylase		E
Transcobalamin 2, TCN2	TCN2	T
Transcription factor 1, hepatic	TCF1	G
Transcription factor 2, hepatic	TCF2	G
Transcription termination factor, RNA polymerase 1	TTF1	G
Transferrin	TF	G



Transferrin receptor	TFRC	G
Transforming growth factor, beta 2	TGFB2	G
Transforming growth factor, beta induced	TGFBI	G
Transforming growth factor, beta receptor 2	TGFBR2	G
Transketolase	TKT	E
Transketolase-like 1	TKTL1	E
Transthyretin	TTR	T
Tubby-like protein 1	TULP1	G
Tuberous sclerosis 1	TSC1	G
Tuberous sclerosis 2	TSC2	G
Tyrosinase	TYR	E
Tyrosinase-related protein 1	TYRP1	E
Tyrosine aminotransferase	TAT	E
Tyrosine hydroxylase	TH	E
Ubiquitin activating enzyme, E1		E
Ubiquitin protein ligase E3A	UBE3A	E
UDP-glucose pyrophosphorylase		E
UDP-glucuronosyltransferase 1	ugt1d, UGT1	E
UDP-glucuronosyltransferase 2	UGT2	E
Uncoupling protein 1		T
Uncoupling protein 3	UCP3	T
Urate oxidase	UOX	E
Ureidopropionase		E
Uridine monophosphate kinase	UMPK	I
Uridine monophosphate synthetase	UMPS	I
Uridinediphosphate(UDP)-galactose-4-epimerase	GALE	E
Uroporphyrinogen decarboxylase	UROD	E
Uteroglobin	UGB	T
Vasoactive intestinal polypeptide	VIP	N
Vasoactive intestinal polypeptide receptor	VIPR	N
Vasoinhibitory peptide		G
Von Hippel-Lindau gene	VHL	G
Werner syndrome helicase	WRN	G
Wolfram syndrome 1 gene	WFS1	S
Xylitol dehydrogenase		E

456. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 455.

457. A set according to claim 455 or 456 in which a minority of said probes for listed genes are absent.

458. A set according to claim 455 or 456 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.
459. A set according to claim 455 or 456 in which a limited number of probes are replaced by probes for non-listed genes.
460. A set of probes for a core group of genes according to any of claims 455 to 459 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.
461. A set according to any of claims 455 to 460 consisting of probes for members of a sub-group of the core group.
462. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.
463. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.
464. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.
465. A set according to claim 462 or 463 in which said substrate is a semiconductor microchip.
466. A set according to any preceding claim for use in a biological assay for detection of said gene variants.
467. A set according to any preceding claim for use in the measurement of differential gene expression levels.
468. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
469. A medical device including a set according to any of claims 455 to 467 for use in an array for detection of differential gene expression levels.
470. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 455) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 455 and 457 to 467 and relating the probe hybridisation pattern to said variations.
471. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 456) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 456 to 467 and relating the probe interaction pattern to said variations.
472. Use of a set or device according to any of claims 455 to 467 for the prognosis and management of patients suffering from or at risk of experiencing the symptoms and consequences of endocrine and metabolic dysfunction, damage or disease.
473. Use of a set or device according to any of claims 455 to 467 for predicting likely therapeutic response and adverse events following therapeutic intervention.

474. Use of a set or device according to any of claims 455 to 467 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
475. Use of a set or device according to any of claims 455 to 467 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
476. Use of a set or device according to any of claims 455 to 467 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
477. Use of a set or device according to any of claims 455 to 467 for the development of new strategies of therapeutic intervention and in clinical trials.
478. Use of a set or device according to any of claims 455 to 467 for construction of and generation of algorithms for patient and healthcare management.
479. Use of a set or device according to any of claims 455 to 467 for modelling or assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations
480. Use of a set or device according to any of claims 455 to 467 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
481. Use of a set or device according to any of claims 455 to 467 for predicting optimum configuration/management of thereapeutic intervention.
482. A method according to claim 470 or 471 in which the identification of gene variants is indicative of a higher risk of developing the symptoms and consequences of endocrine and metabolic dysfunction, damage or disease, for the patient or individual.
483. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop the symptoms and consequences of endocrine and metabolic dysfunction, damage or disease, which method comprises:
- i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from the symptoms and consequences of endocrine and metabolic dysfunction, damage or disease;
  - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the symptoms and consequences of endocrine and metabolic dysfunction, damage or disease;
  - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 455 to 461;
  - iv) calculating the frequencies of these alleles in the samples from i) and ii);
  - v) comparing the frequencies of these alleles in i) and ii);
  - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing the symptoms and consequences of endocrine and metabolic dysfunction, damage or disease.
484. A method for assessing whether a given subject will be at risk of developing symptoms, which comprises comparing said subject's genotype with a model generated by the method of claim 483.
485. A method according to any of claims 470, 471, 483 and 484 wherein at least one step is computer-controlled.

486. An assay suitable for use in a method according to any of claims 470, 471, 483 and 484; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 455 to 461 in a biological sample.
487. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing the symptoms and consequences of endocrine and metabolic dysfunction, damage or disease; said kit comprising:
- i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 455 or 457 to 461 in a sample of human DNA;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing the symptoms and consequences of endocrine and metabolic dysfunction, damage or disease.
488. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing the symptoms and consequences of endocrine and metabolic dysfunction, damage or disease; said kit comprising:
- i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core group of genes as defined in any of claims 456 to 461 in an expressed-protein-containing human sample;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing the symptoms and consequences of endocrine and metabolic dysfunction, damage or disease.
489. A set of probes according to claim 455, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.
490. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to headaches; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

#### KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME  
 T TRANSPORT & STORAGE  
 S STRUCTURAL  
 I IMMUNITY  
 N NERVOUS TRANSMISSION  
 G GROWTH & DIFFERENTIATION

#### HEADACHE GENE LIST

HUGO gene  
symbol

Protein  
function

Acetylcholinesterase	ACHE	E
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
Angiopietin 1	ANGPT1	G
Angiopietin 2	ANGPT2	G
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensin receptor 1	AGTR1	T
Angiotensin receptor 2	AGTR2	T
Angiotensinogen	AGT	E
Arginase	ARG1	E
Arginine vasopressin	AVP	N
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G
Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
Calcitonin/Calcitonin gene-related peptide	CALCA	N
alpha		
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha-1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha-1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha-1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha-1E (CACNL1A6)	CACNA1E	N
Calcium channel, voltage-dependent, Alpha-2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N
Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	CACNA1A	N
Calcium channel, voltage-dependent, T-type		N

Calnexin	CANX	G
Cannabinoid receptor	CNR1	N
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Catechol-O-methyltransferase	COMT	E
Choline acetyltransferase	CHAT	E
Cyclic AMP-dependent protein kinase	PKA	E
Cyclic nucleotide phosphodiesterase 1B	PDE1B	E
Cyclic nucleotide phosphodiesterase 1B1	PDE1B1	E
Cyclic nucleotide phosphodiesterase 2A3	PDE2A3	E
Cyclic nucleotide phosphodiesterase 3A	PDE3A	E
Cyclic nucleotide phosphodiesterase 3B	PDE3B	E
Cyclic nucleotide phosphodiesterase 4A	PDE4A	E
Cyclic nucleotide phosphodiesterase 4C	PDE4C	E
Cyclic nucleotide phosphodiesterase 5A	PDE5A	E
Cyclic nucleotide phosphodiesterase 6A	PDE6A	E
Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E
Cyclic nucleotide phosphodiesterase 8	PDE8	E
Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
CYP11A1	CYP11A1	E
CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E
CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E
CYP2C18	CYP2C18	E
CYP2C19	CYP2C19	E
CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E
CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E

CYP2F1	CYP2F1	E
CYP2J2	CYP2J2	E
CYP3A3	CYP3A3	E
CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystathionase	CTH	E
Cystathione beta synthase	CBS	E
Cytidine deaminase	CDA	E
Cytidine-5-prime-triphosphate synthetase	CTPS	E
Cytochrome a		E
Cytochrome c		E
Cytochrome c oxidase, MTCO		E
Cytokine-suppressive antiinflammatory drug-binding protein 1	CSBP1	I
Cytokine-suppressive antiinflammatory drug-binding protein 2	CSBP2	I
Dopamine beta hydroxylase	DBH	E
Dopamine receptors D1	DRD1	N
Dopamine receptors D2	DRD2	N
Dopamine receptors D3	DRD3	N
Dopamine receptors D4	DRD4	N
Dopamine receptors D5	DRD5	N
Dystonia 9	CSE	S
Endothelin 1	EDN1	N
Endothelin 2	EDN2	N
Endothelin 3	EDN3	N
Endothelin converting enzyme	ECE1	N
Endothelin receptor type A	EDNRA	N
Endothelin receptor type B	EDNRB	N
Enolase	ENO1	E
Epidermal growth factor	EGF	G
Epidermal growth factor receptor	EGFR	G
Erythropoietin receptor	EPOR	I
Glutathione	GSH	T
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
Glycerol kinase	GK	E
Glycinamide ribonucleotide (GAR)	GART	E

transformylase		
Hexosaminidase B	HEXB	E
Histamine receptors, H1		N
Histamine receptors, H2		N
Histamine receptors, H3		N
Hypoxia inducible factor 1	HIF1A	E
Hypoxia inducible factor 2		E
Insulin	INS	G
Insulin receptor	INSR	G
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
IP3 kinase		E
Marenostrin	MEFV	T
Methylmalonyl-CoA mutase	MUT	E
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Myogenic factor 3	MYF3	G
Myogenic factor 4	MYF4	G
Myogenic factor 5	MYF5	G
NADH dehydrogenase		E
NADPH-dependent cytochrome P450 reductase	POR	E
Neurokinin A	NKNA	N
Neurokinin B	NKNB	N
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Nitric oxide synthase 1, NOS1	NOS1	E
Nitric oxide synthase 2, NOS2	NOS2	E
Nitric oxide synthase 3, NOS3	NOS3	E
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C alpha		I
Phospholipase C beta		I
Phospholipase C delta	PLCD1	I
Phospholipase C epsilon		I



Phospholipase C gamma	PLCG1	I
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium voltage-gated channel E1	KCNE1	N
Potassium voltage-gated channel Q1	KCNQ1	N
Proopiomelanocortin	POMC	N
RIGUI	RIGUI	G
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage-gated, type 1, beta	SCN1B	N
polypeptide		
Solute carrier family 5, member 3	SLC5A3	T
Solute carrier family 6 (GAMMA-AMINO-BUTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Substance P		N
Tyrosine hydroxylase	TH	E
UDP-glucuronosyltransferase 1	ugt1d, UGT1	E
UDP-glucuronosyltransferase 2	UGT2	E
Vasoactive intestinal polypeptide	VIP	N
Vasoactive intestinal polypeptide receptor	VIPR	N

491. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 490.

492. A set according to claim 490 or 491 in which a minority of said probes for listed genes are absent.
493. A set according to claim 490 or 491 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.
494. A set according to claim 490 or 491 in which a limited number of probes are replaced by probes for non-listed genes.
495. A set of probes for a core group of genes according to any of claims 490 to 494 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.
496. A set according to any of claims 490 to 495 consisting of probes for members of a sub-group of the core group.
497. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.
498. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.
499. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.
500. A set according to claim 497 or 498 in which said substrate is a semiconductor microchip.
501. A set according to any preceding claim for use in a biological assay for detection of said gene variants.
502. A set according to any preceding claim for use in the measurement of differential gene expression levels.
503. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
504. A medical device including a set according to any of claims 490 to 502 for use in an array for detection of differential gene expression levels.
505. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 490) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 490 and 492 to 502 and relating the probe hybridisation pattern to said variations.
506. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 491) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 491 to 502 and relating the probe interaction pattern to said variations.
507. Use of a set or device according to any of claims 490 to 502 for the prognosis and management of patients suffering from or at risk of developing a headache.

508. Use of a set or device according to any of claims 490 to 502 for predicting likely therapeutic response and adverse events following therapeutic intervention.
509. Use of a set or device according to any of claims 490 to 502 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
510. Use of a set or device according to any of claims 490 to 502 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
511. Use of a set or device according to any of claims 490 to 502 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
512. Use of a set or device according to any of claims 490 to 502 for the development of new strategies of therapeutic intervention and in clinical trials.
513. Use of a set or device according to any of claims 490 to 502 for construction of and generation of algorithms for patient and healthcare management.
514. Use of a set or device according to any of claims 490 to 502 for modelling or assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations.
515. Use of a set or device according to any of claims 490 to 502 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
516. Use of a set or device according to any of claims 490 to 502 for predicting optimum configuration/management of therapeutic intervention.
517. A method according to claim 505 or 506 in which the identification of gene variants is indicative of a higher risk of developing a headache for the patient or individual.
518. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop a headache, which method comprises:
- i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from headaches;
  - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the headaches;
  - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 490 to 496;
  - iv) calculating the frequencies of these alleles in the samples from i) and ii);
  - v) comparing the frequencies of these alleles in i) and ii);
  - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing a headache.
519. A method for assessing whether a given subject will be at risk of developing symptoms, which comprises comparing said subject's genotype with a model generated by the method of claim 518.
520. A method according to any of claims 505, 506, 518 and 519 wherein at least one step is computer-controlled.
521. An assay suitable for use in a method according to any of claims 505, 506, 518 and 519; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 490 to 496 in a biological sample.

522. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing a headache; said kit comprising:
- i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 490 or 492 to 496 in a sample of human DNA;
  - ii) reagents for use in the detection process
  - ii) readout indicating the probability of a patient or individual developing a headache.
523. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing a headache; said kit comprising:
- i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core group of genes as defined in any of claims 491 to 496 in an expressed-protein-containing human sample;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing a headache.
524. A set of probes according to claim 490, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.
525. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to sexual dysfunction; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

#### KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME  
 T TRANSPORT & STORAGE  
 S STRUCTURAL  
 I IMMUNITY  
 N NERVOUS TRANSMISSION  
 G GROWTH & DIFFERENTIATION

#### SEXUAL DYSFUNCTION GENE LIST

	HUGO gene symbol	Protein function
11beta hydroxysteroid dehydrogenase 2	HSD11B2	E
Acetylcholinesterase	ACHE	E
Activin		G
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E

Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
Adrenoleukodystrophy gene	ALD	E
alpha thalassemia gene	ATRX	N
Androgen binding protein	ABP	T
Angiopoietin 1	ANGPT1	G
Angiopoietin 2	ANGPT2	G
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensin receptor 1	AGTR1	T
Angiotensin receptor 2	AGTR2	T
Angiotensinogen	AGT	E
Anti-Mullerian hormone	AMH	G
Anti-Mullerian hormone type 2 receptor	AMHR2	G
Arginase	ARG1	E
Arginine vasopressin	AVP	N
Arginine vasopressin receptor 1A	AVPR1A	N
Arginine vasopressin receptor 1B	AVPR1B	N
Arginine vasopressin receptor 2	AVPR2	N
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G
Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
Autoimmune regulator, AIRE	AIRE	I
BCL2-associated X protein	BAX	G
Bloom syndrome protein	BLM	G
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha-1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha-1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha-1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha-1E (CACNL1A6)	CACNA1E	N
Calcium channel, voltage-dependent, Alpha-2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N

Calcium channel, voltage-dependent, T-type		N
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Catechol-O-methyltransferase	COMT	E
Choline acetyltransferase	CHAT	E
Cyclic AMP response element modulator	CREM	G
Cyclic AMP-dependent protein kinase	PKA	E
Cyclic nucleotide phosphodiesterase 1B	PDE1B	E
Cyclic nucleotide phosphodiesterase 1B1	PDE1B1	E
Cyclic nucleotide phosphodiesterase 2A3	PDE2A3	E
Cyclic nucleotide phosphodiesterase 3A	PDE3A	E
Cyclic nucleotide phosphodiesterase 3B	PDE3B	E
Cyclic nucleotide phosphodiesterase 4A	PDE4A	E
Cyclic nucleotide phosphodiesterase 4C	PDE4C	E
Cyclic nucleotide phosphodiesterase 5A	PDE5A	E
Cyclic nucleotide phosphodiesterase 6A	PDE6A	E
Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E
Cyclic nucleotide phosphodiesterase 8	PDE8	E
Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
CYP11A1	CYP11A1	E
CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E
CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E
CYP2C18	CYP2C18	E
CYP2C19	CYP2C19	E
CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E
CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E

CYP2F1	CYP2F1	E
CYP2J2	CYP2J2	E
CYP3A3	CYP3A3	E
CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystathionase	CTH	E
Cystathione beta synthase	CBS	E
Cytidine deaminase	CDA	E
Cytidine-5-prime-triphosphate synthetase	CTPS	E
Cytochrome a		E
Cytochrome c		E
Cytochrome c oxidase, MTCO		E
Cytokine-suppressive antiinflammatory drug-binding protein 1	CSBP1	I
Cytokine-suppressive antiinflammatory drug-binding protein 2	CSBP2	I
DAX1 nuclear receptor	DAX1	I
Deleted in azoospermia	DAZ	G
Diaphanous 2	DIAPH2	N
Disrupted meiotic cDNA 1, homolog	DMC1	G
Dopamine beta hydroxylase	DBH	E
Dopamine receptors D1	DRD1	N
Dopamine receptors D2	DRD2	N
Dopamine receptors D3	DRD3	N
Dopamine receptors D4	DRD4	N
Dopamine receptors D5	DRD5	N
Electron-transferring-flavoprotein alpha	ETFA	T
Electron-transferring-flavoprotein beta	ETFB	T
Electron-transferring flavoprotein dehydrogenase	ETFDH	E
Endometrial bleeding-associated factor	EBAF	G
Endothelin 1	EDN1	N
Endothelin 2	EDN2	N
Endothelin 3	EDN3	N
Endothelin converting enzyme	ECE1	N
Endothelin receptor type A	EDNRA	N
Endothelin receptor type B	EDNRB	N
Enolase	ENO1	E
Enoyl CoA isomerase		E

Enterokinase	PRSS7, ENTK	E
Epidermal growth factor	EGF	G
Epidermal growth factor receptor	EGFR	G
Facio-genital dysplasia	FGD1, FGDY	T
Factor XIII A & B	F13A & F13B	I
Fanconi anemia, complementation group A	FANCA	T
Fertilin protein	FTNB	G
Flightless-II, Drosophila homolog of	FLII	G
Folic acid receptor	FOLR	G
Glutathione	GSH	T
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
Glycerol kinase	GK	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
Glycogen phosphorylase	PYGL	E
Gonadotropin releasing hormone	GNRH	G
Gonadotropin releasing hormone receptor	GNRHR	G
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 1, GNAI1	GNAI1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 2, GNAI2	GNAI2	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 3, GNAI3	GNAI3	N
Hexosaminidase B	HEXB	E
Holoprosencephaly 1	HPE1	G
Holoprosencephaly 2	HPE2	G
Holoprosencephaly 3	HPE3	G
Holoprosencephaly 4	HPE4	G
Human placental lactogen	CSH1	G
Inhibin, alpha	INHA	G
Inhibin, beta A	INHBA	G
Inhibin, beta B	INHBB	G
Inhibin, beta C	INHBC	G
Insulin	INS	G
Insulin receptor	INSR	G
IP3 kinase		E
Kallman syndrome gene 1	KAL1	G
Laminin 5, alpha 3	LAMA3	G
Laminin 5, beta 3	LAMB3	G
Laminin receptor 1	LAMR1	G
Long QT-type 2 potassium channels	LQT2, KCNH2	T
Luteinizing hormone, beta chain	LHB	G
MAD (mothers against decapentaplegic, Drosophila) homologue 2	MADH2	G
Methylmalonyl-CoA mutase	MUT	E
Monoamine oxidase A	MAOA	E



Monoamine oxidase B	MAOB	E
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
NADPH-dependent cytochrome P450 reductase	POR	E
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Nitric oxide synthase 1, NOS1	NOS1	E
Nitric oxide synthase 2, NOS2	NOS2	E
Nitric oxide synthase 3, NOS3	NOS3	E
Oncogene ELK1	ELK1	G
Oncogene ELK2	ELK2	G
Paired box homeotic gene 3	PAX3	G
Patched (Drosophila) homolog, PTCH	PTCH	G
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium inwardly-rectifying channel J11	KCNJ11	N
Potassium voltage-gated channel A1	KCNA1	N
Potassium voltage-gated channel E1	KCNE1	N
Potassium voltage-gated channel Q1	KCNQ1	N
Potassium voltage-gated channel Q2	KCNQ2	N
Potassium voltage-gated channel Q3	KCNQ3	N
Progesterone receptor (RU486 binding receptor)	PGR	G
Proopiomelanocortin	POMC	N
Prostasin, PRSS8	PRSS8	E
Ribosomal protein S4, X-linked	RPS4X	E
RIGUI	RIGUI	G
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N

Sodium channel, voltage gated, type V, alpha polypeptide	SCN5A	N
Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Solute carrier family 6 (GAMMA-AMINOBTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Sperm protamine P1	PRM1	G
Sperm protamine P2	PRM2	G
T-BOX 3	TBX3	G
Testis-specific protein Y	TSPY	G
Tyrosine hydroxylase	TH	E
UDP-glucuronosyltransferase 1	ugt1d, UGT1	E
UDP-glucuronosyltransferase 2	UGT2	E
Vasoactive intestinal polypeptide	VIP	N
Vasoactive intestinal polypeptide receptor	VIPR	N
Zona pellucida glycoprotein 1	ZP1	G
Zona pellucida glycoprotein 2	ZP2	G
Zona pellucida glycoprotein 3	ZP3	G
Zona pellucida receptor tyrosine kinase	ZRK	G
Zonadhesin	ZAN	G

526.A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 525.

527.A set according to claim 525 or 526 in which a minority of said probes for listed genes are absent.

528.A set according to claim 525 or 526 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.

529.A set according to claim 525 or 526 in which a limited number of probes are replaced by probes for non-listed genes.

530.A set of probes for a core group of genes according to any of claims 525 to 529 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.

531. A set according to any of claims 525 to 530 consisting of probes for members of a sub-group of the core group.
532. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.
533. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.
534. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.
535. A set according to claim 532 or 533 in which said substrate is a semiconductor microchip.
536. A set according to any preceding claim for use in a biological assay for detection of said gene variants.
537. A set according to any preceding claim for use in the measurement of differential gene expression levels.
538. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
539. A medical device including a set according to any of claims 525 to 537 for use in an array for detection of differential gene expression levels.
540. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 525) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 525 and 527 to 537 and relating the probe hybridisation pattern to said variations.
541. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 526) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 526 to 537 and relating the probe interaction pattern to said variations.
542. Use of a set or device according to any of claims 525 to 537 for the prognosis and management of patients suffering from or at risk of developing sexual dysfunction.
543. Use of a set or device according to any of claims 525 to 537 for predicting likely therapeutic response and adverse events following therapeutic intervention.
544. Use of a set or device according to any of claims 525 to 537 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
545. Use of a set or device according to any of claims 525 to 537 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
546. Use of a set or device according to any of claims 525 to 537 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
547. Use of a set or device according to any of claims 525 to 537 for the development of new strategies of therapeutic intervention and in clinical trials.
548. Use of a set or device according to any of claims 525 to 537 for construction

- of and generation of algorithms for patient and healthcare management.
549. Use of a set or device according to any of claims 525 to 537 for modelling or assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations
550. Use of a set or device according to any of claims 525 to 537 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
551. Use of a set or device according to any of claims 525 to 537 for predicting optimum configuration/management of therapeutic intervention.
552. A method according to claim 540 or 541 in which the identification of gene variants is indicative of a higher risk of developing sexual dysfunction for the patient or individual.
553. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop sexual dysfunction which method comprises:
- i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from sexual dysfunction;
  - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the sexual dysfunction;
  - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 525 to 531;
  - iv) calculating the frequencies of these alleles in the samples from i) and ii);
  - v) comparing the frequencies of these alleles in i) and ii);
  - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing sexual dysfunction.
554. A method for assessing whether a given subject will be at risk of developing symptoms, which comprises comparing said subject's genotype with a model generated by the method of claim 553.
555. A method according to any of claims 540, 541, 553 and 554 wherein at least one step is computer-controlled.
556. An assay suitable for use in a method according to any of claims 540, 541, 553 and 554; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 525 to 531 in a biological sample.
557. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing sexual dysfunction; said kit comprising:
- i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 525 or 527 to 531 in a sample of human DNA;
  - ii) reagents for use in the detection process
  - iii) readout indicating the probability of a patient or individual developing sexual dysfunction.
558. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing sexual dysfunction; said kit comprising:
- i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core

group of genes as defined in any of claims 526 to 531 in an expressed-protein-containing human sample;

- ii) reagents for use in the detection process
- iii) readout indicating the probability of a patient or individual developing sexual dysfunction.

559. A set of probes according to claim 525, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.

# 1/2 SCHIZOPHRENIA

Nonadherence is common, especially if patients do not collaborate in their choice of treatment

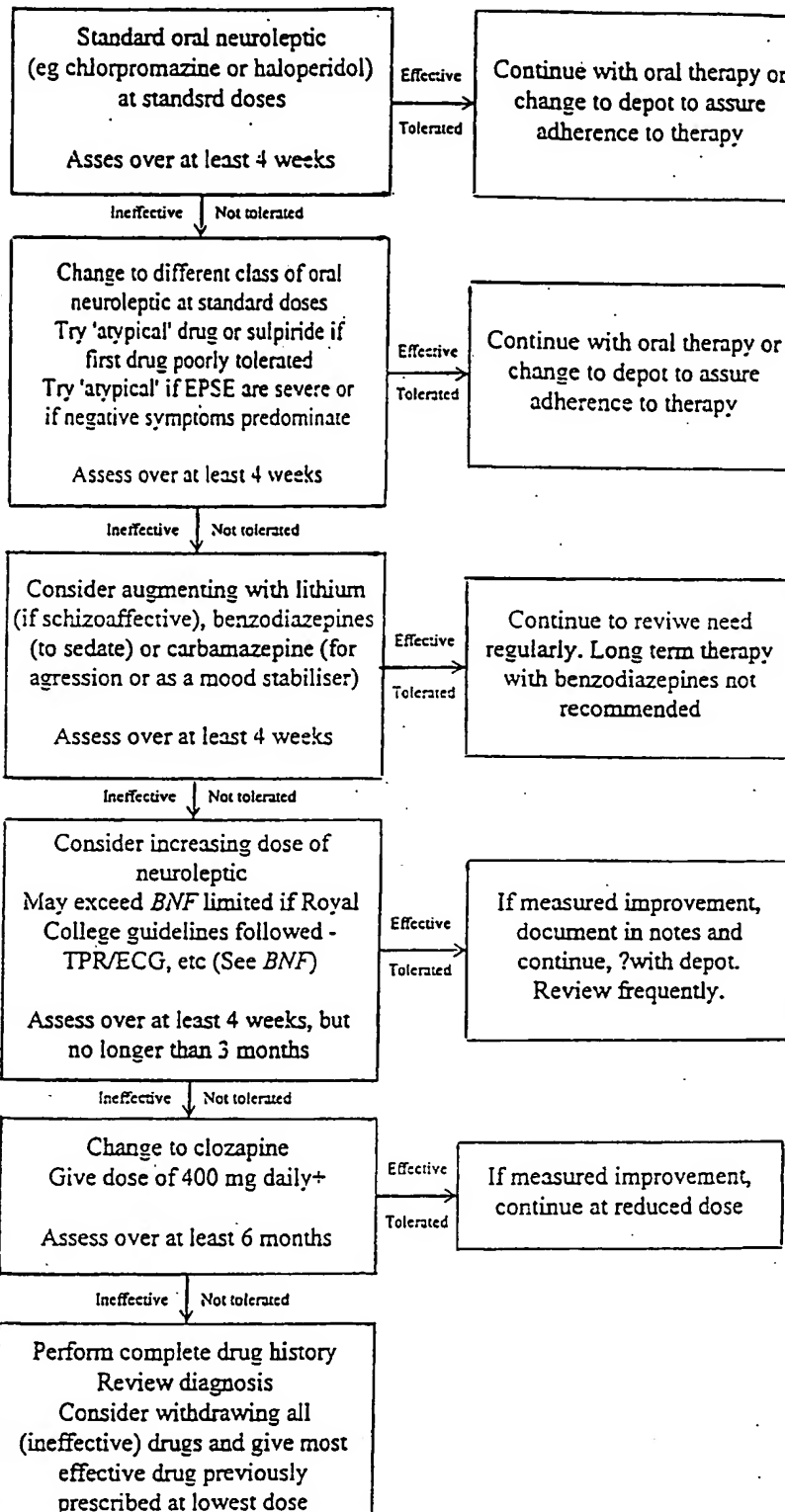
Assess efficiency and tolerance with recognised rating scales, eg BPRS, PANSS, ESRS, LUNBERS

Avoid neuroleptic, polypharmacy - oral + depot are rarely necessary

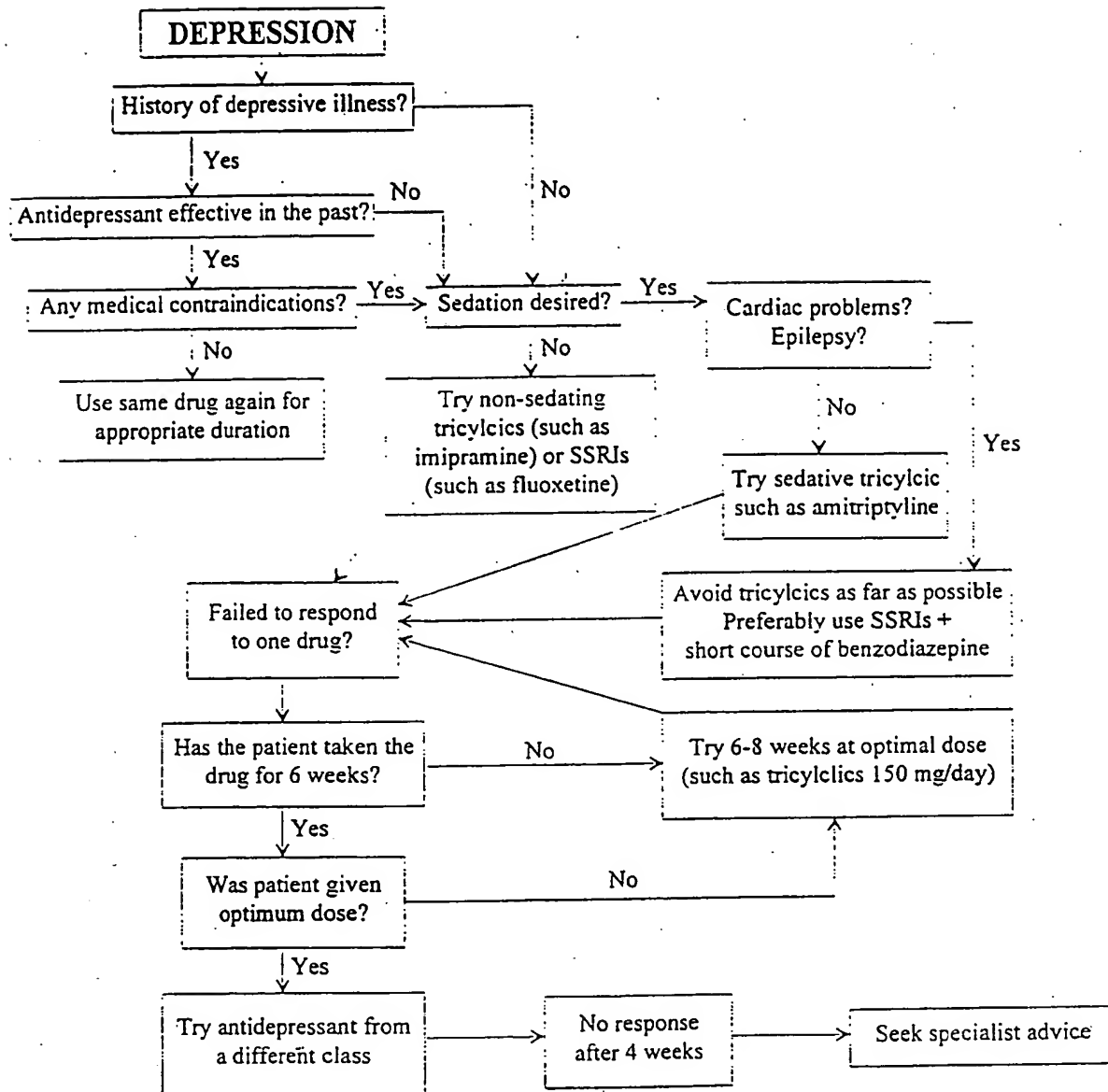
Consider early use of short term clonazepam if sedation is required in acute psychosis

Few data to support the use of high-dose neuroleptics. Do not exceed recommended dose for 'atypical' drugs

Some support for the use of clozapine plasma levels - aim for a pre-dose level of 350 mcg per litre



# DEPRESSION



while the airway responsiveness in our  $F_2$  progeny (C57BL/6J x A/J) was unimodally and approximately normally distributed. Several factors may account for these differences, including methods of assessment of airway reactivity, differences in mouse age<sup>35</sup>, the susceptibility of acetylcholine to cleavage, housing conditions and viral antigen free (VAF) status. We minimized these factors by studying only male mice of fixed age housed under barrier conditions, and provoked airway responses with an acetylcholine congener not subject to cleavage by acetylcholinesterase.

In humans, it has been postulated that asthma can be inherited as a mendelian dominant<sup>6</sup> (with variable penetrance) or polygenic<sup>36</sup> trait. Longo *et al.* examined the airway responsiveness to carbachol in healthy parents of asthmatic children<sup>6</sup>. The presence of a bimodal distribution in airway responsiveness in the parent population and the observation that 85% of the couples who had an asthmatic child had at least one or two parents with normal airway responsiveness suggested that bronchial reactivity was inherited as an autosomal dominant trait with incomplete penetrance. Although the possibility of ascertainment bias exists, the presence of a bimodal distribution of bronchial responsiveness in nonasthmatic parents of asthmatics suggests a single genetic locus. Our results are similar in that about half of the observed variance in airway responsiveness could be attributed to environmental factors. Townley *et al.*<sup>36</sup> demonstrated a unimodal distribution of airway responsiveness in normal subjects from control, nonasthmatic, non-allergic families and confirmed a bimodal distribution of MCh responses in families with and without asthma. The heterogeneity of bronchial responses were significantly affected by both the age and sex of subjects<sup>36</sup>. Their conclusion was that the bronchial response to MCh is not due to segregation at a single locus.

Given that airway responsiveness to MCh did not segregate as a single locus, we used the approach of Sewell Wright<sup>37</sup> to estimate the segregation index or number of loci responsible for regulating airway responsiveness. This approach makes the assumption that all loci make equal contributions to the expression of the phenotypes in question. Estimation of the number of loci responsible for the phenotypic effect of interest was calculated according to these principles<sup>37</sup>. This analysis suggests the involvement of two loci to regulate airway hyperresponsiveness in the mouse. Since we have already identified 3 loci and yet 74% of the genetic variance remains unaccounted for, it seems likely that the simple assumptions made by Wright (such as equal and additive phenotypic effects of the loci) do not apply, and that loci interact in a more complicated manner to cause airway hyperresponsiveness. Indeed, we found evidence of epistatic interaction among the three loci.

Given the biological significance of the candidate genes linked to *Bhr1*, *Bhr2* and *Bhr3* to asthma, one can now evaluate the importance of these specific genes in asthma or asthma models. This approach could consist of determining if there exist differences in sequence or expression of candidate genes in "asthma" and "nonasthma-like" murine strains. Construction of congenic animals by the directed backcross of (C57BL/6J x A/J)  $F_1$  progeny onto a C57BL/6J background may also be useful for evaluating the relative importance of the various candidate genes to the asthma-like phenotype.

## Methods

**Animals.** Male, viral-antigen-free (VAF) C57BL/6J, ( $n=40$ ), A/J, ( $n=42$ ), (C57BL/6J x A/J)  $F_1$  hybrids, ( $n=40$ ), (A/J x C57BL/6J)  $F_1$  hybrids, ( $n=40$ ), (C57BL/6J x A/J)  $F_2$  intercross mice, ( $n=40$ ), 19 strains of AXB and BXA recombinant inbred mice, ( $n=155$ ) and [(C57BL/6J x A/J) x C57BL/6J] backcross mice ( $n=321$ ) 5–6 weeks of age were purchased from Jackson Laboratory, Bar Harbor, Maine. To minimize environmental effects, VAF mice were housed in isolation cages under VAF conditions. The mice were acclimatized for 10–14 days and allowed free access to commercial pelleted mouse feed and water, which were autoclaved to ensure sterility. In addition, blood from sentinel animals was routinely tested to ensure their VAF status. Animals were studied at eight weeks of age.

**Phenotype analysis.** Each mouse was anaesthetized with an intraperitoneal injection of pentobarbital sodium (70–80 mg kg<sup>-1</sup>), (Anthony Products Company, Arcadia, CA). When an acceptable stage of surgical anaesthesia was reached, the metal portion of a 19-gauge tubing adapter was inserted into the trachea and secured in place. An internal jugular vein was cannulated with a saline-filled Silastic catheter (0.06 cm OD, 6–8 cm in length, <0.005 ml vol) attached to a 0.1-ml Hamilton microsyringe (Hamilton Company, Reno, Nevada) and used to administer methacholine (MCh; Acetyl- $\beta$ -methylcholine chloride, Sigma). A thoracotomy was performed so that pleural pressure would equal body surface pressure. The tracheostomy tube was passed through a hole in the plethysmograph chamber and connected via three ports of a four-way connector to a rodent ventilator (Harvard Apparatus, Division of Ealing Scientific, Natick, MA). The ventilator was set to provide 150 breaths/min with tidal volumes of 5–6  $\mu$ l g<sup>-1</sup> and a positive end-expiratory pressure of 3–4 cm H<sub>2</sub>O. These settings provide eupneic ventilation in mice over the weight range studied<sup>38</sup>. Mice were placed in a sealed constant mass plethysmograph consisting of a 1-l bottle insulated from the ambient environment by 1/2-inch foam padding and containing copper mesh to maintain isothermal conditions. The pressure difference between the plethysmograph chamber and the reference chamber was detected with a transducer (Celesco model LCVR 0-2 cm H<sub>2</sub>O, Canoga Park, CA). Changes in lung volume were determined from measured changes in plethysmograph pressure. An electrical signal proportional to flow was obtained by electrical differentiation of the volume signal. The delay between the volume and flow signals was < 0.5 ms. Transpulmonary pressure was measured as the pressure difference between the pressure at the airway opening, measured from the 4th port of the 4-way connector, and the pressure in the plethysmograph itself. The plethysmograph system has been shown to be without significant amplitude distortion or phase shift up to 30 Hz.

Pulmonary resistance ( $R_L$ ) was determined with the use of signals derived from transpulmonary pressure and lung volume<sup>39</sup>. Dose-response curves to methacholine were obtained by administering sequentially increasing doses of MCh (33  $\mu$ g kg<sup>-1</sup> to 3300  $\mu$ g kg<sup>-1</sup>) in a 20- to 35- $\mu$ l volume. The volume of fluid injected with each dose produced no measurable physiological effects. The peak response to each dose was obtained by making 6 serial measurements within the first minute following injection of MCh. Each measurement was calculated via a cross-correlation technique<sup>39</sup> from a number of breaths recorded during the measurement interval. Because the pulmonary response to MCh peaks later and dissipates more slowly with increasing doses, the interval for each of the measurements was increased from 4 s (10 breaths) to 10 s (25 breaths) as the MCh dose was increased. Furthermore, enough time was allowed to elapse between MCh doses such that pre-dose measurements of  $R_L$  returned to within 10% of the value obtained before the preceding dose of MCh. A large breath (three times tidal volume) was administered to standardize volume history prior to each dose of MCh. Pulmonary responses were recorded on a dedicated microcomputer. Each animal's dose-response curve was log-transformed and then subjected to regression analysis to calculate the dose required for a two fold increase in  $R_L$  ( $ED_{200}R_L$ ). The transformation of the phenotype data to a log scale is necessary to normalize the distributions in order to construct genetic maps via the MAPMAKER-QTL computer package<sup>40</sup>.

**DNA preparation.** A midline abdominal incision was performed and the animal killed by exsanguination under surgical anesthesia. Both kidneys were carefully removed, snap frozen in liquid nitrogen



and subsequently stored in a -80 °F freezer. Purified genomic DNA was obtained from one of the kidneys using a DNA extraction kit (Stratagene). Upon completion of the protocol, spectrophotometric readings were taken to verify the purity of the samples (A260/A280) and quantitate (A260 nm) the DNA concentration.

**Genotype analysis.** To genotype backcross progeny for SSLPs, PCR reactions were performed with one radioactively labelled primer and one unlabelled primer and the products were visualized upon autoradiography of polyacrylamide gels. Primers were end-labelled with [ $\gamma$ -<sup>32</sup>P] ATP (Easytides, DuPont) according to standard protocols<sup>41</sup>. A 20 ng aliquot of genomic DNA was amplified in a 10  $\mu$ l PCR reaction using AmpliTaq DNA polymerase (Perkin-Elmer Cetus) according to manufacturer's specifications. The primer concentrations were 75 nM end-labelled forward primer, and 75 nM unlabelled reverse primer. The reactions were overlaid with 40  $\mu$ l of light mineral oil (Sigma). Reactions were amplified on a TC1600 thermal cycler (Intelligent Automation Systems, Cambridge, MA) using the following protocol: 30 cycles of 92 °C for 30 s, 55 °C for 30 s, and 72 °C for 30 s. Gels and autoradiography were as previously described<sup>42</sup>. In total, we used 157 SSR markers that covered the genome at an average spacing of 9 cM (available upon request).

**Linkage analysis and QTL mapping.** The genetic map was constructed by using the MAPMAKER computer package, as described<sup>40</sup>. QTL analysis was performed on the log-transformed phenotype using the MAPMAKER-QTL computer package as described<sup>40</sup>. Statistical analysis. Computations were performed with the

Statview® 4.0 (Abacus Concepts, Berkeley, CA, USA) and JMP® 3.0 (SAS Institute Inc., Cary, NC, USA) statistical packages. A Kruskal-Wallis one-way ANOVA by ranks and a Wald-Wolfowitz unpaired t-test were used for the comparison of the pre-infusion baseline and log ED<sub>50</sub> results in the different parental strains and crosses. A Mann-Whitney U test was used to compare mean dose-response curves for R<sub>1</sub> between A/J and C57BL/6J mice. Where appropriate, a Shapiro-Wilk W test was used to assess normality. Standard analysis-of-variance (ANOVA), including cross-terms for two-way and three-way interactions, were used to evaluate possible interactions between candidate QTL loci (*Bhr1*, *Bhr2*, and *Bhr3*) and mouse airway responsiveness phenotype. Results are expressed as means  $\pm$  standard deviation, unless otherwise stated, were considered statistically significant at the  $P < 0.05$  level.

# Acknowledgements

We thank G. Farino, L. Wangchuk, D. Tsering, K. Norbu for technical assistance; T. Haynes (Jackson Lab) for her assistance and K. Brady, O. Iakoubova and A. De Sanctis for reviewing the manuscript. This work was supported by the Medical Research Council of Canada, and grants from the National Institute of Health (HL36110) and the National Center for Human Genome Research. G.T.D.S. is a recipient of a Medical Research Council of Canada Award.

Received 30 June; accepted 10 August 1995.

- Hirshman, C.A., Downes, H. & Veith, L. Airway responses in offspring of dogs with and without airway hyperactivity. *J. appl. Physiol.* **56**, 1272-1277 (1984).
- Konno, S. *et al.* Bronchial reactivity to methacholine and serotonin in six inbred mouse strains. *Aerugi* **42**, 42-47 (1993).
- Levitt, R.C. & Mitzner, W. Expression of airway hyperactivity to acetylcholine as a simple autosomal recessive trait in mice. *FASEB J.* **2**, 2605-2608 (1988).
- Levitt, R.C. & Mitzner, W. Autosomal recessive inheritance of airway hyperactivity to 5-hydroxytryptamine. *J. appl. Physiol.* **67**, 1125-1132 (1989).
- Guidelines for the diagnosis and management of asthma. National Heart, Lung, and Blood Institute. National Asthma Education Program. Expert Panel Report. *J. Allergy clin. Immunol.* **88**, 425-534 (1991).
- Longo, G., Strinati, R., Poli, F. & Fumi, F. Genetic factors in nonspecific bronchial hyperactivity. An epidemiologic study. *Am. J. Dis. Child.* **141**, 331-334 (1987).
- Nieminen, M.M. Unimodal distribution of bronchial hyperresponsiveness to methacholine in asthmatic patients. *Chest* **102**, 1537-1543 (1992).
- Lander, E.S. & Botstein, D. Mapping mendelian factors underlying quantitative traits using RFLP linkage maps. *Genetics* **121**, 185-199 (1989).
- Broide, D.H. *et al.* Cytokines in symptomatic asthma airways. *J. Allergy clin. Immunol.* **89**, 958-967 (1992).
- Sim, T.C., Hilsmeier, K.A., Reece, L.M., Grant, J.A. & Alam, R. Interleukin-1 receptor antagonist protein inhibits the synthesis of IgE and proinflammatory cytokines by allergen-stimulated mononuclear cells. *Am. J. Respir. Cell. Mol. Biol.* **11**, 473-479 (1994).
- Tsukagoshi, H., Sakamoto, T., Wenbing, X., Barnes, P.J. & Chung, K.F. Effect of interleukin-1b on airway hyperresponsiveness and inflammation in sensitized and nonsensitized Brown-Norway rats. *J. Allergy clin. Immunol.* **93**, 464-9 (1994).
- Gavett, S.H. & Wills-Karp, M. Elevated lung G protein levels and muscarinic receptor affinity in a mouse model of airway hyperactivity. *Am. J. Physiol.* **265**, L493-500 (1993).
- Mikami, H. *et al.* Characteristics of two lines of guinea pigs (BHS and BHR) differing in bronchial sensitivity to acetylcholine and histamine exposure. *Jikken Dobutsu* **40**, 453-460 (1991).
- Barnes, P.J. Cytokines as mediators of chronic asthma. *Am. J. resp. crit. Care Med.* **150**, S42-S49 (1994).
- Aubert, J.D. *et al.* Platelet-derived growth factor and its receptor in lungs from patients with asthma and chronic airflow obstruction. *Am. J. Physiol.* **266**, L655-663 (1994).
- Gordon, J.R., Burd, P.R. & Galli, S.J. Mast cells as a source of multifunctional cytokines. *Immunol. Today* **11**, 458-464 (1990).
- Wasserman, S.I. Mast cell biology. *J. Allergy clin. Immunol.* **86**, 590-593 (1990).
- Pui, C.H. Serum interleukin-2 receptor: clinical and biological implications. *Leukemia* **3**, 323-327 (1989).
- Hashimoto, S. *et al.* Elevation of soluble IL-2 receptor and IL-4, and nonelevation of IFN- $\gamma$  in sera from patients with allergic asthma. *Ann. Allergy* **71**, 455-458 (1993).
- Park, C.S. *et al.* Soluble interleukin-2 receptor and cellular profiles in bronchoalveolar lavage fluid from patients with bronchial asthma. *J. Allergy clin. Immunol.* **91**, 623-633 (1993).
- Matsumoto, K., Taki, F., Miura, M., Matsuzaki, M. & Takagi, K. Serum levels of soluble IL-2R, IL-4, and soluble Fc epsilon RI in adult bronchial asthma. *Chest* **105**, 681-686 (1994).
- Corrigan, C.J., Hartnell, A. & Kay, A.B. T lymphocyte activation in acute severe asthma. *Lancet* **1**, 1129-1132 (1988).
- Van Bever, H.P. *et al.* Effect of a bronchial provocation test with house-dust mite on blood eosinophilia, eosinophil cationic protein, soluble interleukin-2 receptor, and interleukin-6 in asthmatic children. *Allergy* **48**, 443-449 (1993).
- Sekizawa, K., Caughey, G.H., Lazarus, S.C., Gold, W.M. & Nadel, J.A. Mast cell tryptase causes airway smooth muscle hyperresponsiveness in dogs. *J. clin. Invest.* **83**, 175-179 (1989).
- Wenzel, S.E., Fowler, A.A.D. & Schwartz, L.B. Activation of pulmonary mast cells by bronchoalveolar allergen challenge. In vivo release of histamine and tryptase in atopic subjects with and without asthma. *Am. Rev. respir. Dis.* **137**, 1002-1008 (1988).
- Caughey, G.H., Leidig, F., Viro, N.F. & Nadel, J.A. Substance P and vasoactive intestinal peptide degradation by mast cell tryptase and chymase. *J. Pharmacol. exp. Ther.* **244**, 133-137 (1988).
- Lilly, C.M., Martins, M.A. & Drazen, J.M. Peptidase modulation of vasoactive intestinal peptide pulmonary relaxation in tracheal superfused guinea pig lungs. *J. clin. Invest.* **91**, 235-243 (1993).
- Ruoss, S.J., Hartmann, T. & Caughey, G.H. Mast cell tryptase is a mitogen for cultured fibroblasts. *J. clin. Invest.* **88**, 493-499 (1991).
- Ying, S. *et al.* TNF alpha mRNA expression in allergic inflammation. *Clin. exp. Allergy* **21**, 745-750 (1991).
- Kips, J.C., Tavernier, J. & Pauwels, R.A. Tumor necrosis factor causes bronchial hyperresponsiveness in rats. *Am. Rev. respir. Dis.* **145**, 332-336 (1992).
- Martin, T.R. *et al.* Mast cell activation enhances airway responsiveness to methacholine in the mouse. *J. clin. Invest.* **91**, 1176-1182 (1993).
- Renz, H. *et al.* Aerosolized antigen exposure without adjuvant causes increased IgE production and increased airway responsiveness in the mouse. *J. Allergy clin. Immunol.* **89**, 1127-1138 (1992).
- Garsen, J., Nijkamp, F.P., Van Der Vliet, H. & Van Loveren, H. A role for cellular immunity in the induction of airway hyperresponsiveness induced by small molecular weight compounds. *Toxicol. Lett.* **72**, 151-154 (1994).
- Pauwels, R. *et al.* Genetic factors in non-specific bronchial reactivity in rats. *Eur. J. respir. Dis.* **66**, 98-104 (1985).
- Yarkony, K.A. & Wills-Karp, M. The effects of aging on airway hyperactivity in inbred strains of mice. *Am. Rev. respir. Dis.* **147**, A419 (1993).
- Townley, R.G. *et al.* Segregation analysis of bronchial response to methacholine inhalation challenge in families with and without asthma. *J. Allergy clin. Immunol.* **77**, 101-107 (1986).
- Wright, S. *Evolution and the Genetics of Populations: Genetic and Biometric Foundations.* (University of Chicago Press, Chicago, 1968).
- Martin, T.R., Gerard, N.P., Galli, S.J. & Drazen, J.M. Pulmonary responses to bronchoconstrictor agonists in the mouse. *J. appl. Physiol.* **64**, 2318-2323 (1988).
- Kroeker, J.P. Wiener analysis of nonlinear systems using Poisson-Charlier crosscorrelation. *Biol. Cybernet.* **27**, 221-227 (1977).
- Lander, E.S. *et al.* MAPMAKER: an interactive computer package for constructing primary genetic linkage maps of experimental and natural populations. *Genomics* **1**, 174-181 (1987).
- Sambrook, J., Fritsch, E.F. & Maniatis, T. *Molecular Cloning: A Laboratory Manual.* (Cold Spring Harbor Laboratory, New York, 1989).
- Dietrich, W.F. *et al.* A genetic map of the mouse with 4,006 simple sequence length polymorphisms. *Nature Genet.* **7**, 220-245 (1994).
- Paterson, A.H. *et al.* Mendelian factors underlying quantitative traits in tomato: comparison across species, generations, and environments. *Genetics* **127**, 181-97 (1991).